

# Congenital Heart Disease

## An Approach for Simple and Complex Anomalies



Michael D. Pettersen, MD  
Director, Echocardiography  
Rocky Mountain Hospital for Children  
Denver, CO

## Disclosures

- None

## ASCeXAM

- Contains questions on general congenital heart disease, not “adult” CHD
- Study guide contain all of the information in this talk plus addition topics that will be helpful for the exam
- There have been a few questions on fetal echo which have appeared on the ASCEXAM which are covered in the handout
- Insider information provided in study guide – topics that have appeared on prior exams (last page of study guide)

Which heart defect is more common?

- ★ 1. Tetralogy of Fallot  
2. Transposition of the Great Arteries  
3. Aortic Stenosis  
4. Coarctation of the Aorta  
5. Atrioventricular Canal

What is the most common defect seen with Down syndrome (trisomy 21) ?

1. Patent Ductus Arteriosus
2. Tetralogy of Fallot
3. Aortic Stenosis
- ★4. Ventricular septal defect
5. Coarctation of the Aorta

## Congenital Heart Disease

Spectrum of Congenital Heart Disease - Incidence

- 0.5–0.8% of live births \*
- >30,000 individuals/year in U.S.
- 50% simple shunts (ASD, VSD, PDA)  
20% simple obstruction  
30% complex

\* Excludes MVP (4-6%) & Bicuspid AV (1-2%)

## Congenital Heart Disease

Spectrum of Congenital Heart Disease - Frequency

<b><u>Cardiac Malformation</u></b>	<b><u>% of CHD</u></b>	<b><u>M:F Ratio</u></b>
Ventr. Septal Defect	18-28	1:1
Patent Ductus Arter.	10-18	1:2-3
Tetralogy of Fallot	10-13	1:1
Atrial Septal Defect	7-8	1:2-4
Pulmonary Stenosis	7-8	1:1
Transp. of Grt. Art.	4-8	2-4:1
Coarctation of Aorta	5-7	2-5:1
AV Septal Defect	2-7	1:1
Aortic Stenosis	2-5	4:1
Truncus Arteriosus	1-2	1:1
Tricuspid Atresia	1-2	1:1
Tot. Anom Pulm Veins	1-2	1:1

## Congenital Heart Disease

Common Syndromes/Chromosomal Anomalies

<b><u>Anomaly</u></b>	<b><u>Associations</u></b>
Trisomy 21	VSD, AV Canal
Trisomy 18	VSD, PDA
Trisomy 13	VSD, PDA, Dextrocardia
Turner	Coarctation, AS
Noonan	PS, HCM
Williams	Supravalvar AS, Peripheral PS
Holt-Oram	ASD
Marfan	Aortic root dilation, MVP
DiGeorge	VSD, arch anomalies, TOF

## Congenital Heart Disease

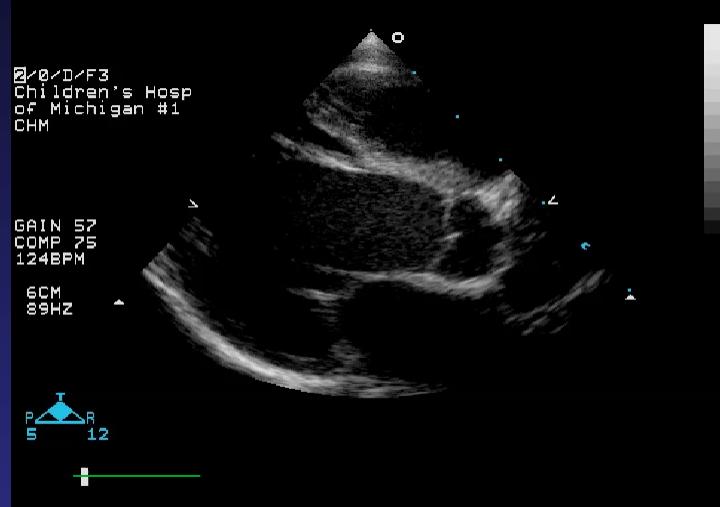
- Currently over 1 million patients over 18 with CHD are alive in this country
  - This increases at 4-5% per year
  - 20,000 cardiac surgical procedures for CHD/year
  - >90% of children with CHD survive to adulthood
  - The majority of adult CHD patients will be post-op
- The ASCEXAM does not cover much post-op disease, but has asked questions about operations

## Congenital Heart Disease

### A Brief History of Operations

Year	Physician	Procedure
1938	Gross	Ligation of PDA
1944	<u>Blalock, Taussig</u>	Syst.-pulm. shunt
1945	Gross, Crafoot	Repair of coarctation
1946	<u>Potts</u>	Aorta to LPA shunt/direct anastomosis
1952	Muller	Pulm. artery band
1953	Gibbon	Repair of ASD
1954	Lillehei	Repair of VSD
1954	<u>Glenn</u>	SVC-PA shunt
1956-7	<u>Mustard/Senning</u>	Atrial correction of TGA
1955	Lillehei, Kirklin	Repair of tetralogy of Fallot
1960	<u>Waterston</u>	Aorta to RPA shunt/direct anastomosis
1964	<u>Rastelli</u>	Conduit replacement of PA
1967	<u>Rashkind</u>	Balloon atrial septostomy
1971	<u>Fontan, Kreutzer</u>	Repair of tricuspid atresia
1976	<u>Jatene</u>	Arterial switch for TGA
1978		Cold blood cardioplegia

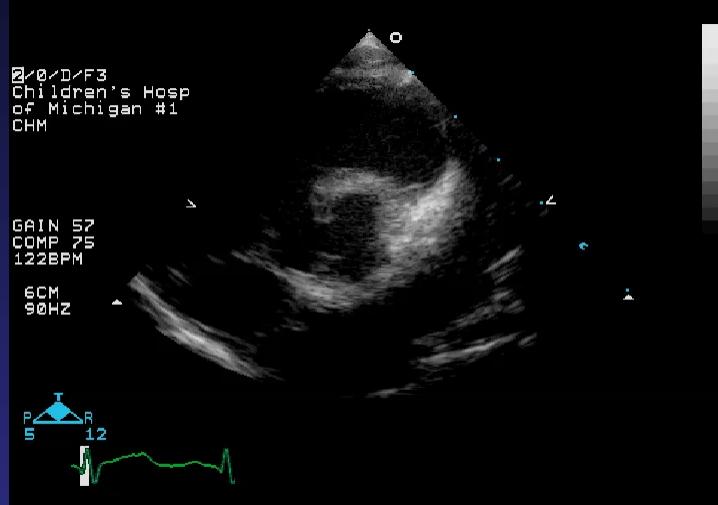
Case 1 - 3 month old with a murmur,  
tachypnea and failure to thrive



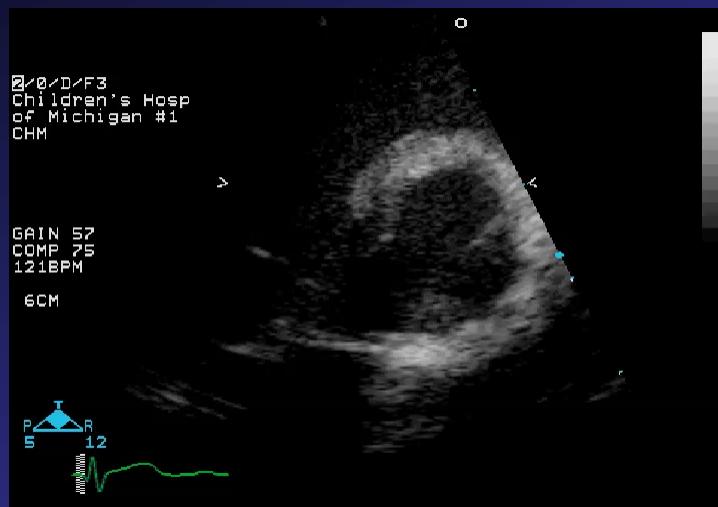
Case 1 - 3 month old with a murmur,  
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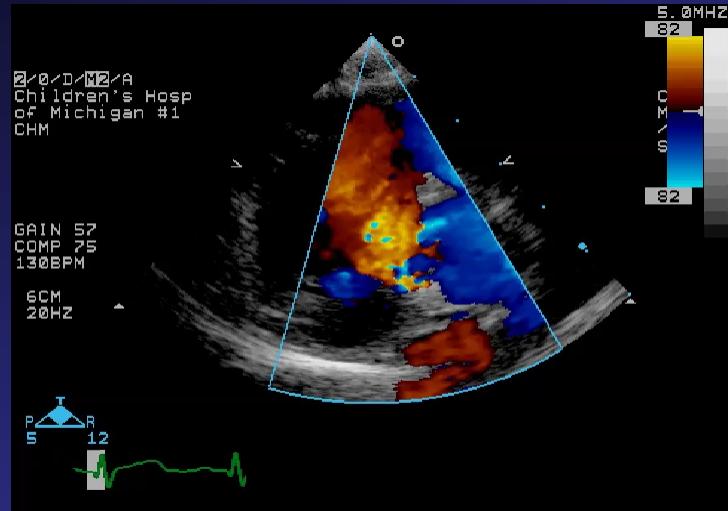
Case 1 - 3 month old with a murmur,  
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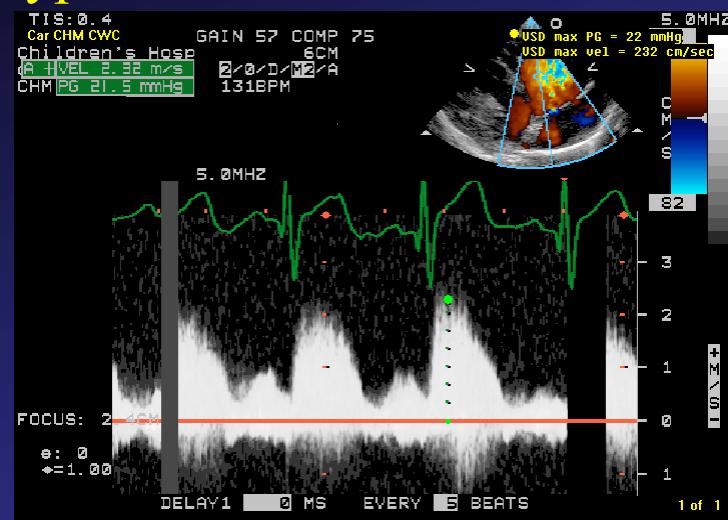
Case 1 - 3 month old with a murmur,  
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## Case 1 - 3 month old with a murmur, tachypnea and failure to thrive



## Case 1 - 3 month old with a murmur, tachypnea and failure to thrive



The defect shown in this example is:

1. Secundum VSD
2. Sinus Venosus VSD
- ★ 3. Perimembranous VSD
4. Inlet VSD
5. Supracristal VSD

An isolated VSD will generally produce enlargement of which chamber(s):

- ★
1. Left atrium, Left ventricle
  2. Right ventricle
  3. Right ventricle, pulmonary artery
  4. Aorta
  5. Right ventricle, right atrium

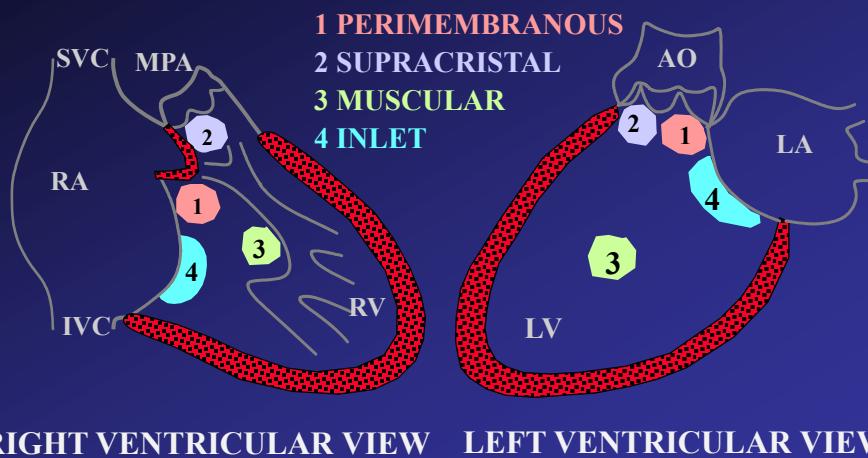
## What is the right ventricular pressure?

1. Normal
2. Supra systemic
- ★ 3. Systemic
4. Can't tell from information given
5. Want to go home now

## Ventricular Septal Defect Clinical

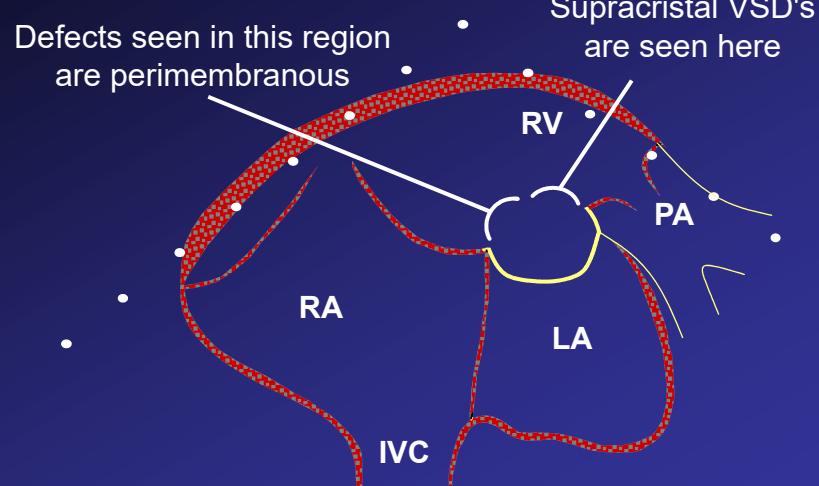
- Most common defect, 25% of CHD
- Shunt flow usually left to right in infants and children
- Symptoms depend on the size of the defect
  - Large - >50% of aortic annulus size
  - Medium - 25-50% of annulus size
  - Small - <25% of annulus size
- Large VSDs result in pulmonary edema
  - tachypnea, poor feeding, failure to thrive in infants
- In un-operated patients with large defects pulmonary vascular disease develops → shunt reversal and cyanosis (Eisenmenger's complex)

# Ventricular Septal Defect Anatomy



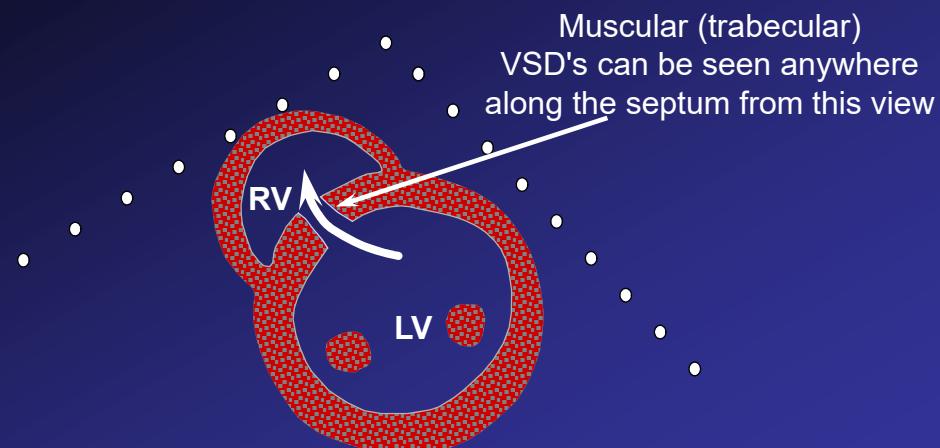
RIGHT VENTRICULAR VIEW    LEFT VENTRICULAR VIEW

## Ventricular Septal Defect Parasternal Short Axis - Base



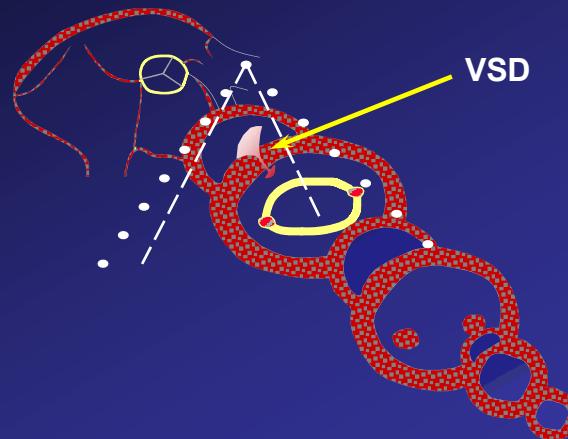
## Ventricular Septal Defect

Parasternal Short-Axis - Mid-Ventriele



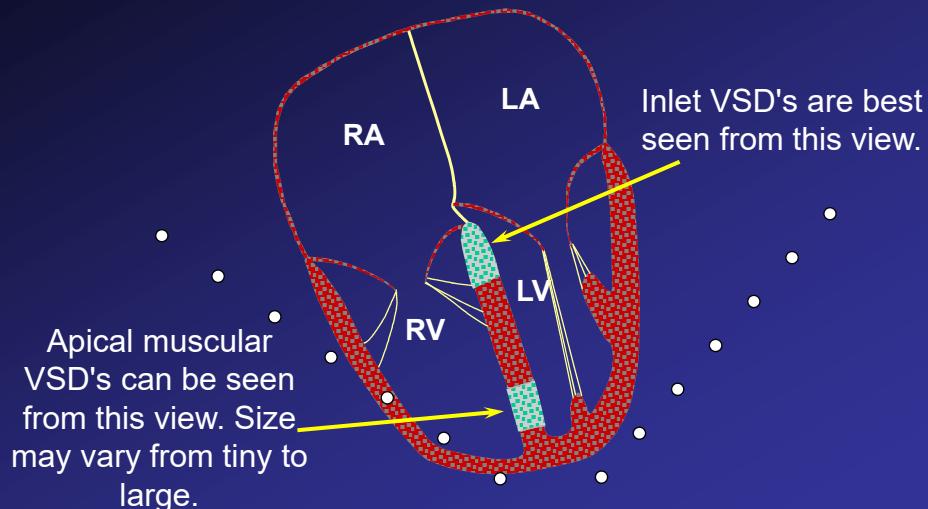
## Ventricular Septal Defect

Parasternal Short-Axis "Sweep"



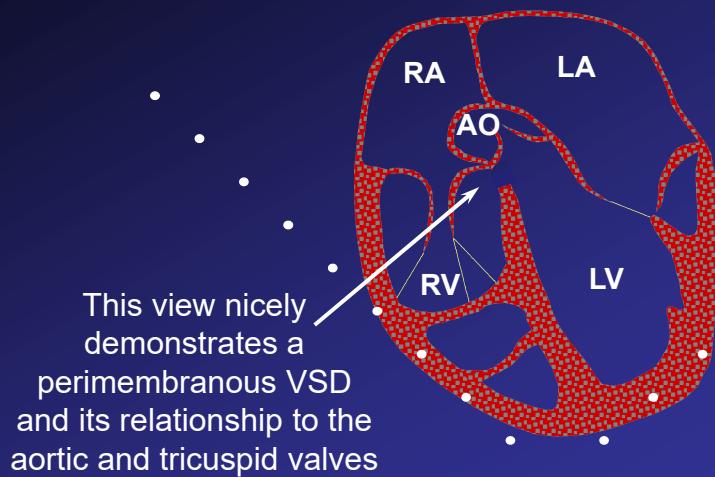
## Ventricular Septal Defect

### Apical Four-Chamber View

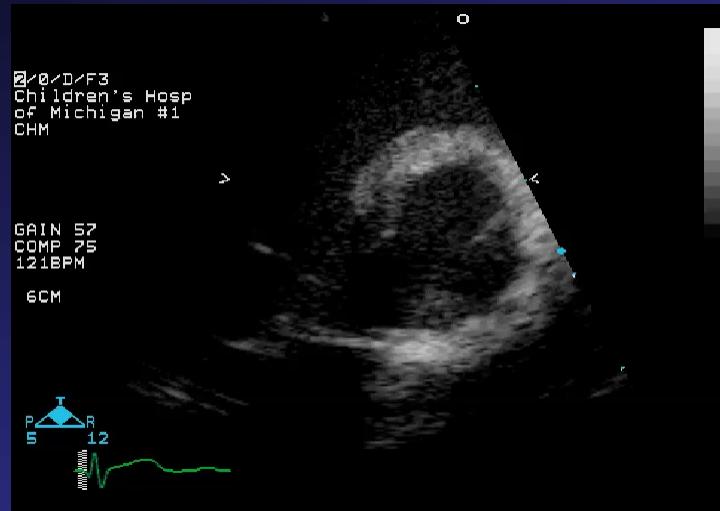


## Ventricular Septal Defect

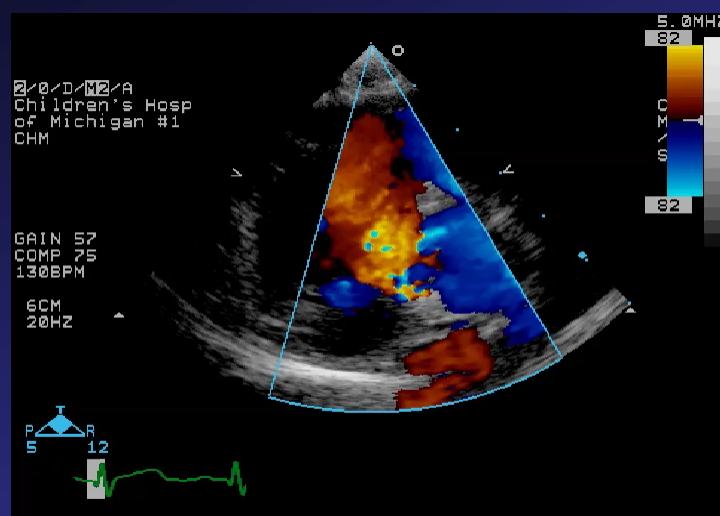
### Apical Five-Chamber View



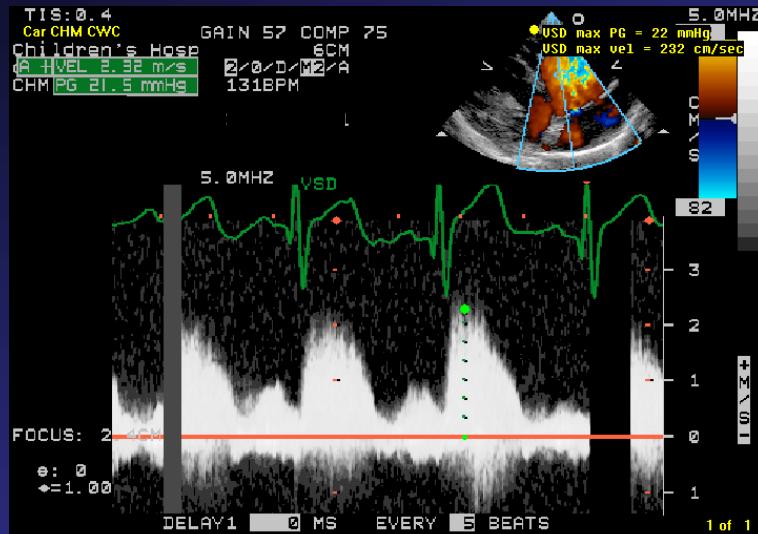
## Case 1- Review



## Case 1- Review

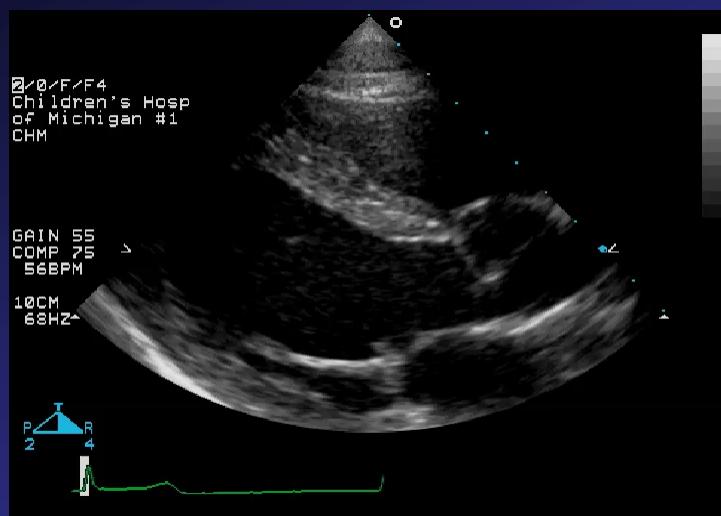


## Case 1- Review



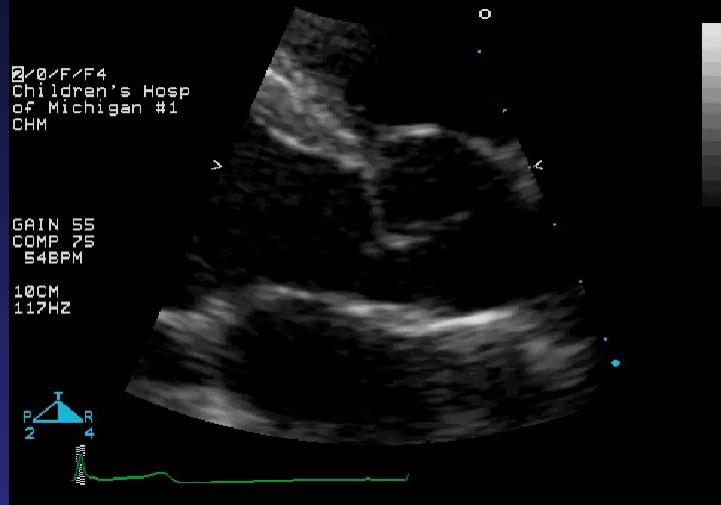
## Ventricular Septal Defect

Case 2 – 8 y.o. with asymptomatic murmur



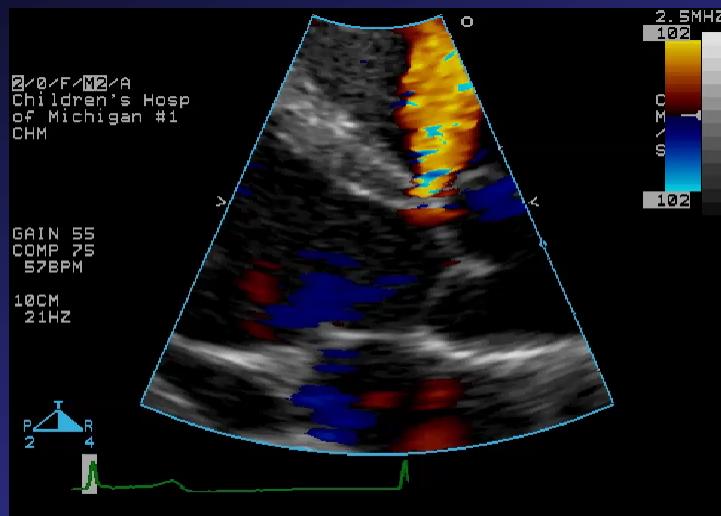
## Ventricular Septal Defect

Case 2 – 8 y.o. with asymptomatic murmur



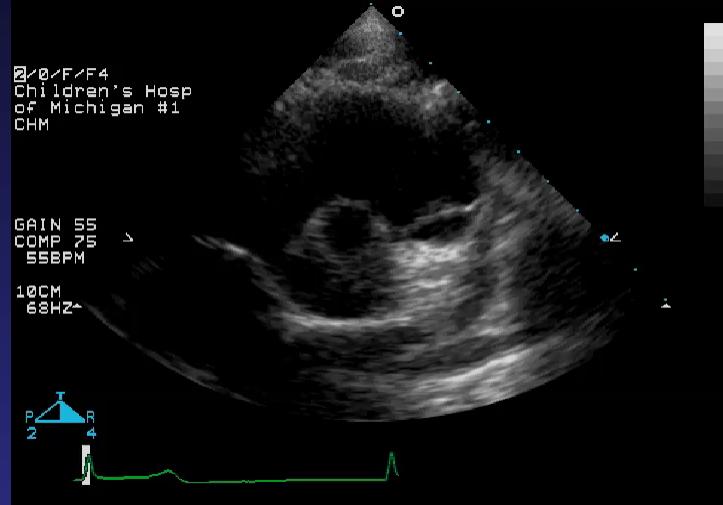
## Ventricular Septal Defect

Case 2 – 8 y.o. with asymptomatic murmur



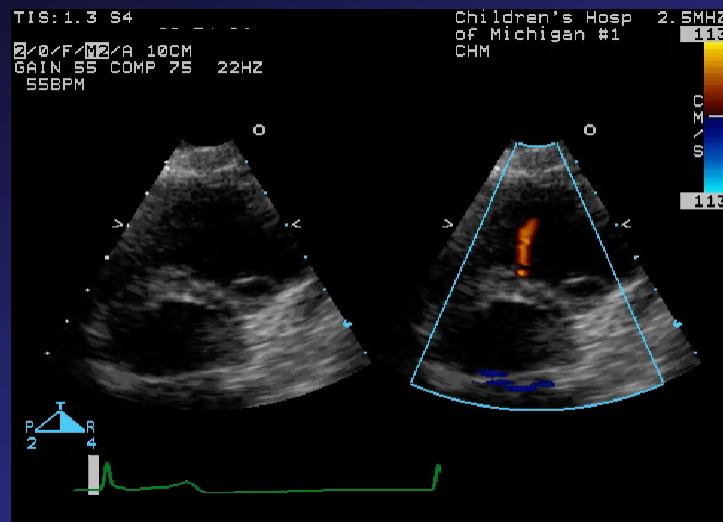
## Ventricular Septal Defect

Case 2 – 8 y.o. with asymptomatic murmur



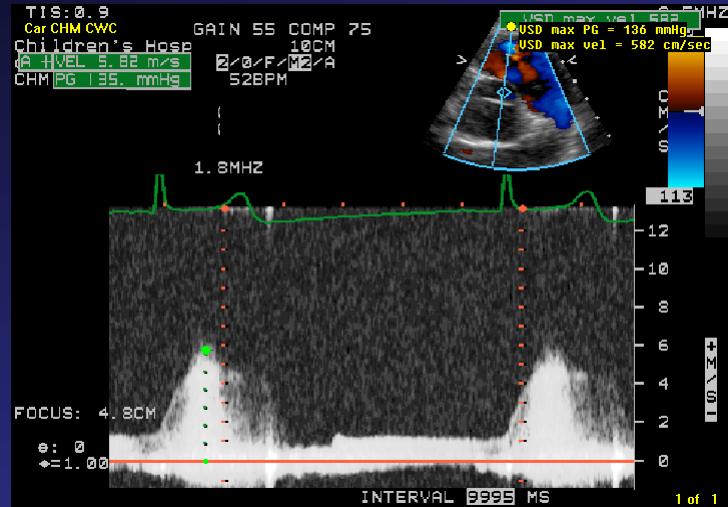
## Ventricular Septal Defect

Case 2 – 8 y.o. with asymptomatic murmur



## Ventricular Septal Defect

Case 2 – 8 y.o. with asymptomatic murmur



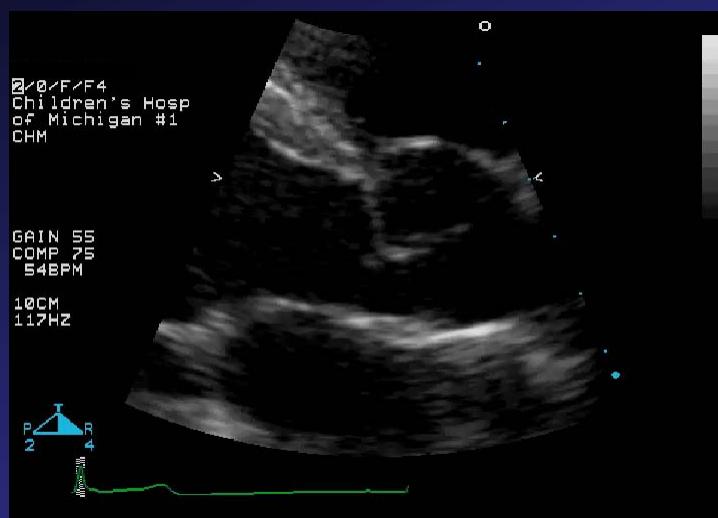
The defect shown in this example is:

1. Secundum VSD
2. Sinus Venosus VSD
3. Perimembranous VSD
4. Inlet VSD
- ★5. Supraventricular VSD

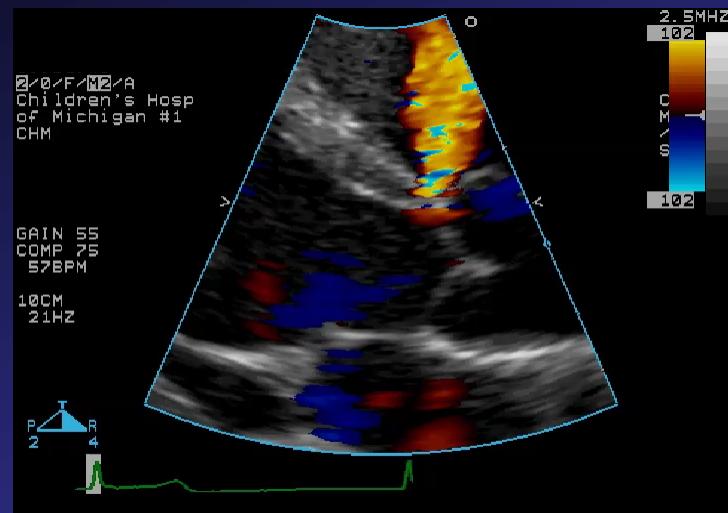
## Question 10 - A common complication of this defect is:

1. Pulmonary valve endocarditis
- ★ 2. Aortic regurgitation
3. Aortic dissection
4. Tricuspid regurgitation
5. Right ventricular enlargement

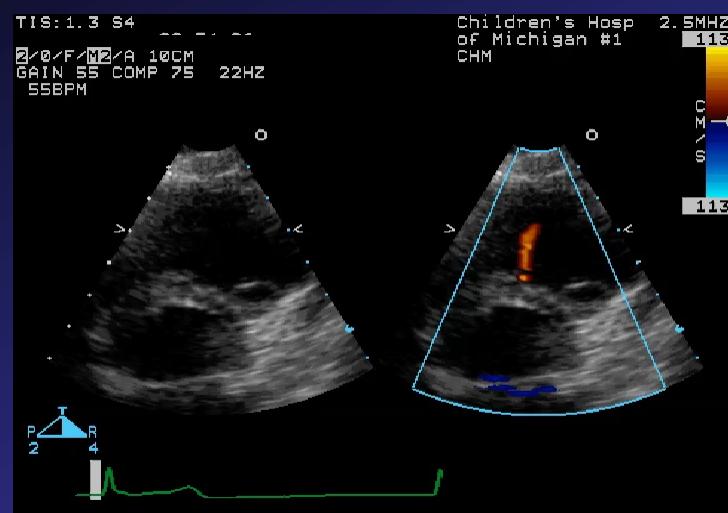
## Case 2- Review



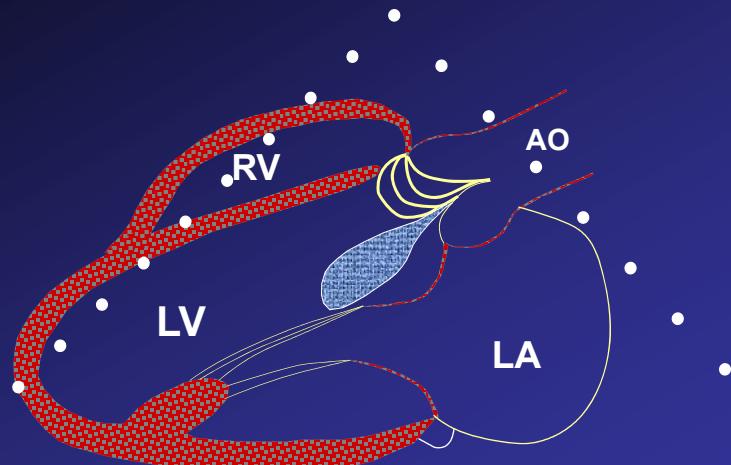
## Case 2- Review



## Case 2- Review



## Supracristal VSD Aortic Cusp Prolapse



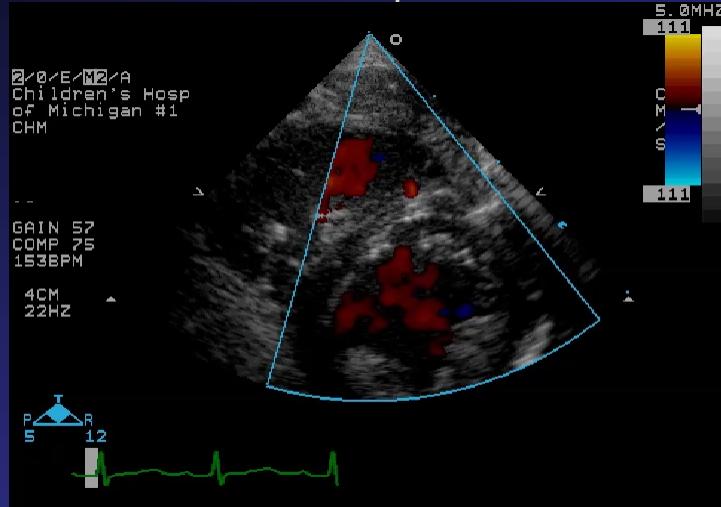
## Ventricular Septal Defect

Case 3 – No questions



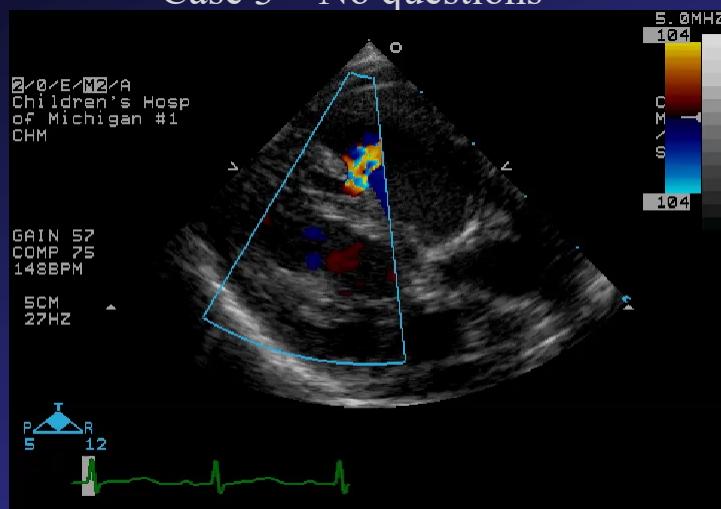
## Ventricular Septal Defect

### Case 3 – No questions



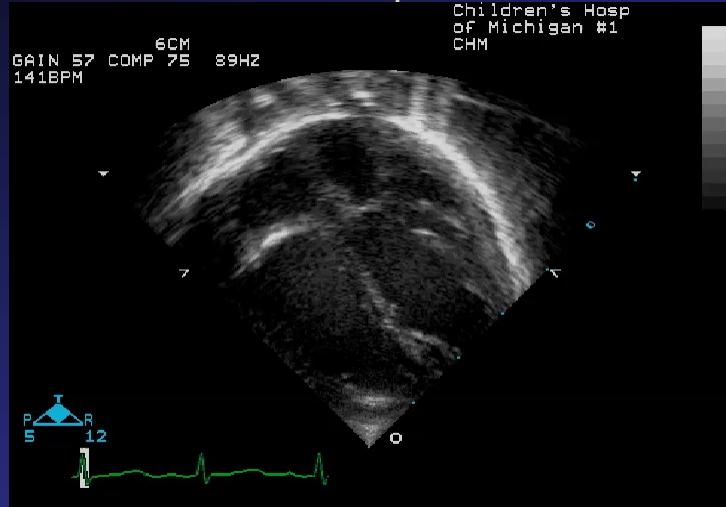
## Ventricular Septal Defect

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## Ventricular Septal Defect

### Case 3 – No questions



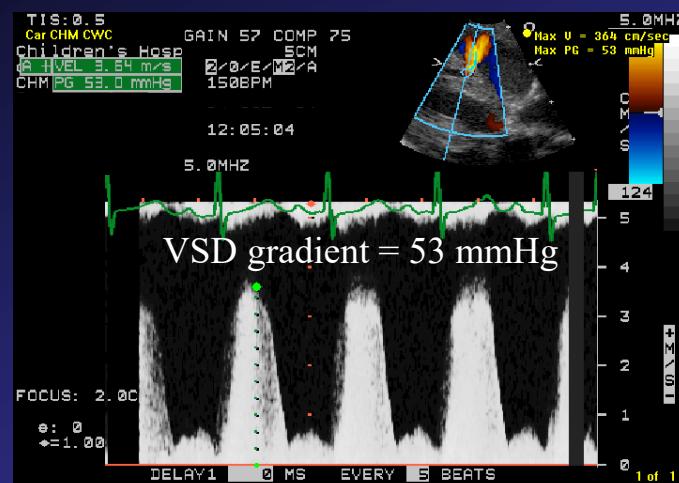
## Ventricular Septal Defect

### Case 3 – No questions



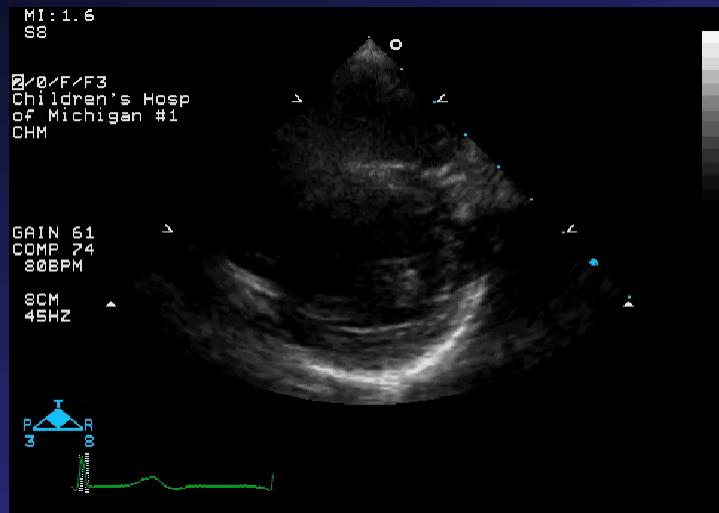
# Ventricular Septal Defect

## Case 3 – No questions

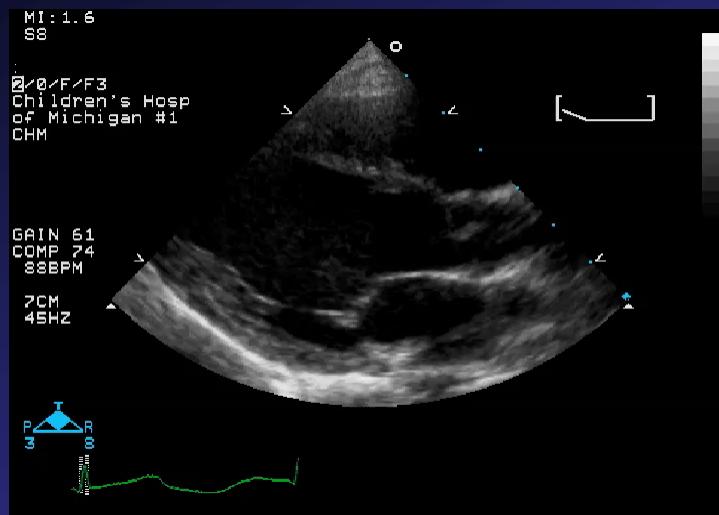


$$\text{BP} = 72/38 \text{ mmHg} \quad \text{RVSP} = 72 - 53 = 19 \text{ mmHg}$$

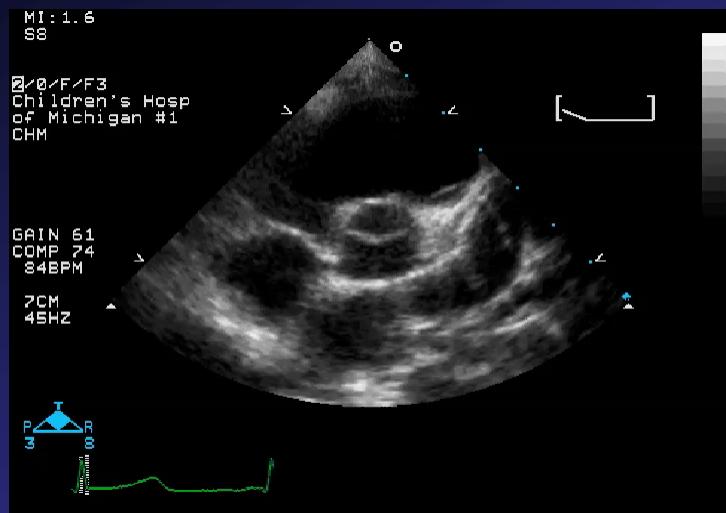
## Case 4 – 6 y.o. with continuous



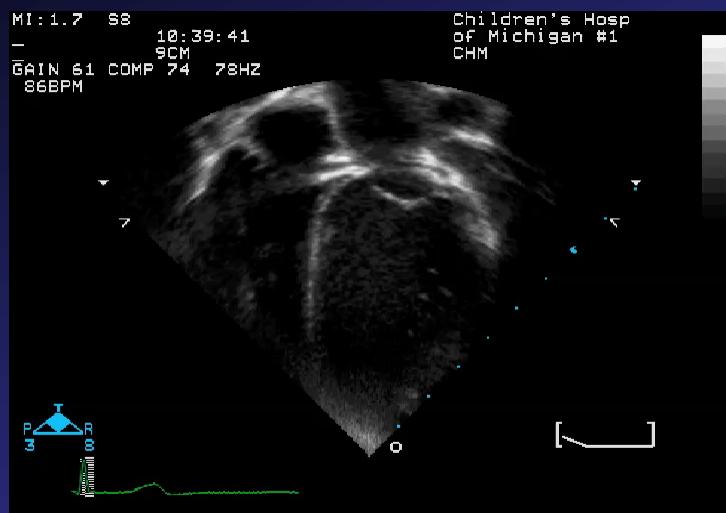
## Case 4 – 6 y.o. with continuous



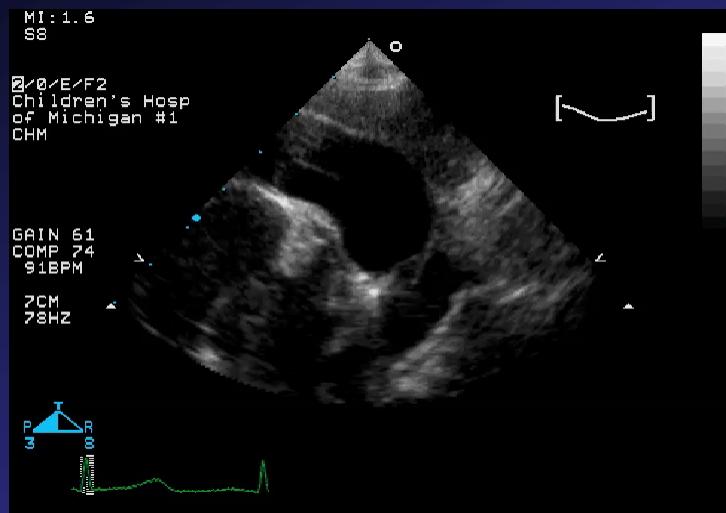
## Case 4 – 6 y.o. with continuous



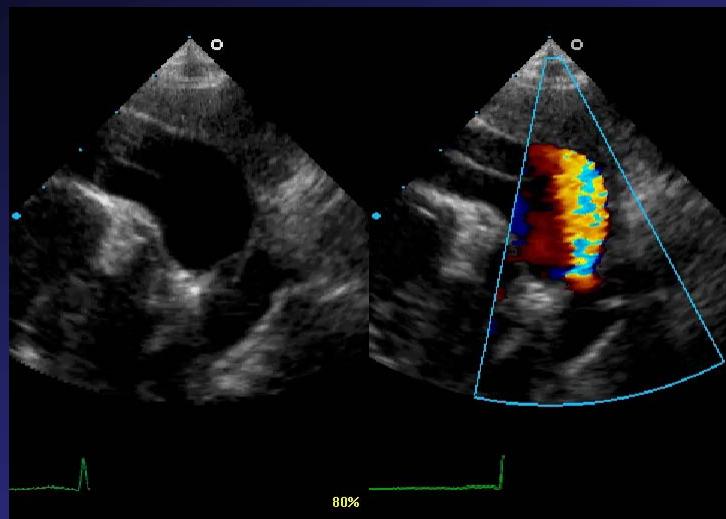
## Case 4 – 6 y.o. with continuous



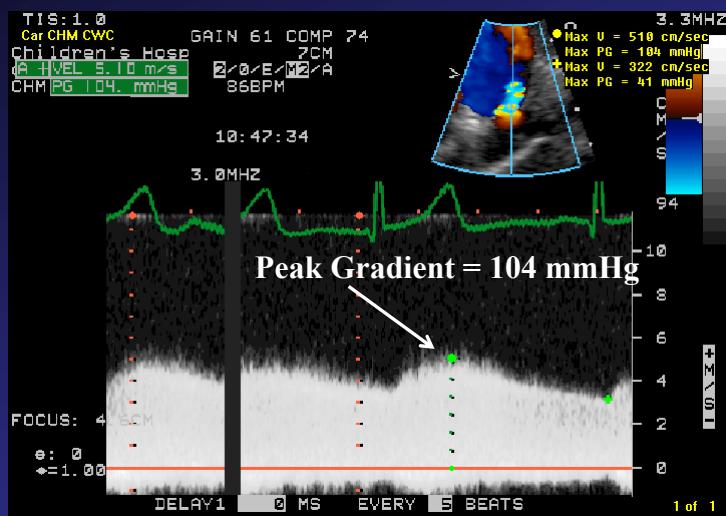
## Case 4 – 6 y.o. with continuous



## Case 4 – 6 y.o. with continuous



## Case 4 – 6 y.o. with continuous murmur



The Doppler tracing in this case implies:

1. Severe pulmonary hypertension
2. Severe systemic hypertension
3. Severe coarctation of the aorta
- ★ 4. Normal pulmonary artery pressure
5. Severe pulmonary artery stenosis

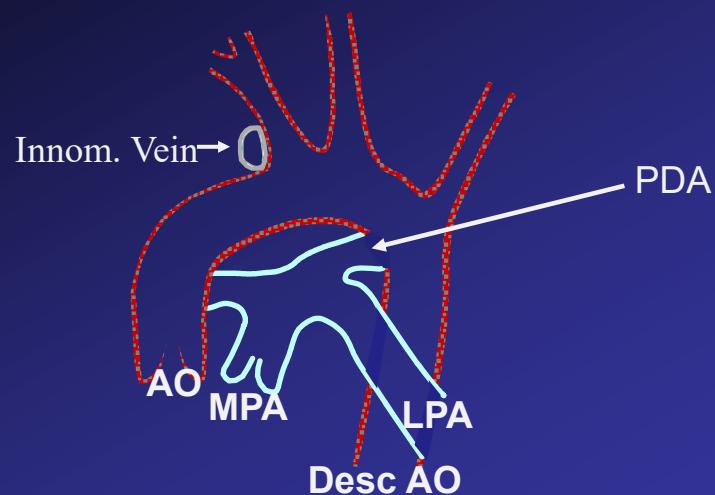
## Patent Ductus Arteriosus

### Clinical

- Continuous murmur in older patients
- Bounding pulses, wide pulse pressure, respiratory symptoms in neonates with a large PDA
- Large PDA will act much like a large VSD, producing pulmonary over-circulation and signs/symptoms of congestive heart failure
- A small PDA is generally hemodynamically insignificant but is at risk for endarteritis

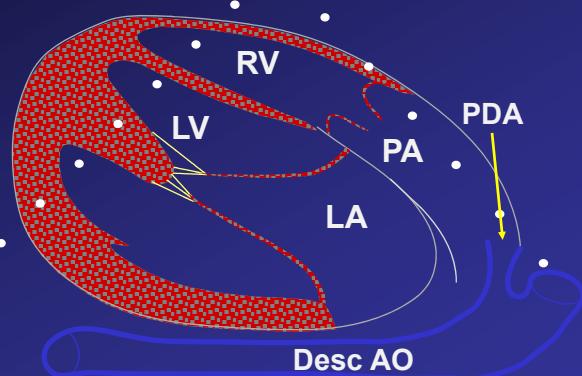
## Patent Ductus Arteriosus

### Anatomy

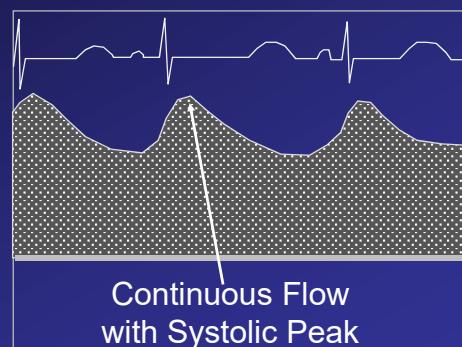
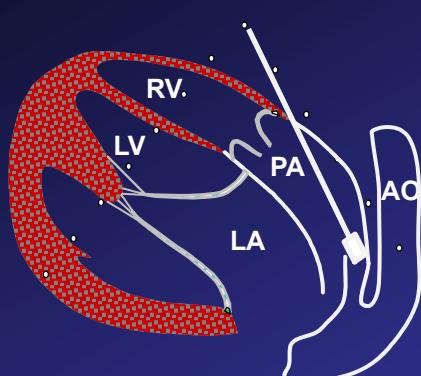


## Patent Ductus Arteriosus

Ductal View  
Parasternal Ductal View  
(High Left Parasternal)



## Patent Ductus Arteriosus Doppler Flow Pattern



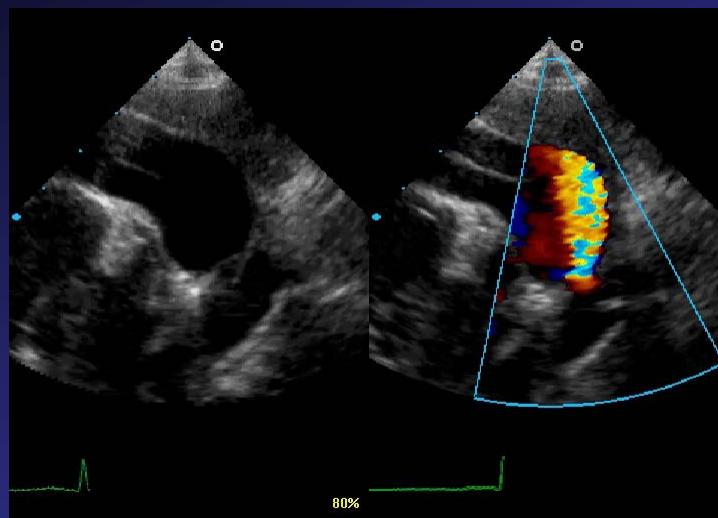
$$\text{Systolic PA pressure} = \text{SBP} - 4V_{\text{PDA}}^2$$

# Patent Ductus Arteriosus

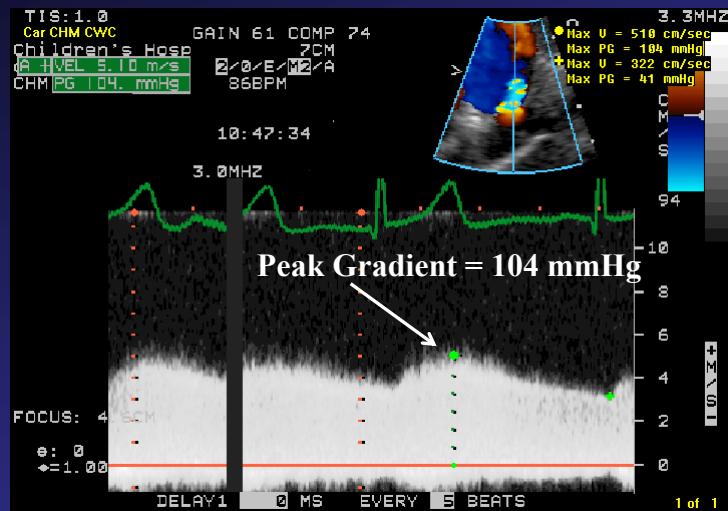
## Clinical management

- Large PDAs in preterm infants
  - Pharmacologic closure – indomethacin or ibuprofen
  - Surgical closure – left lateral thoracotomy
- Small PDA in older infants and children
  - Catheter closure – device or coil

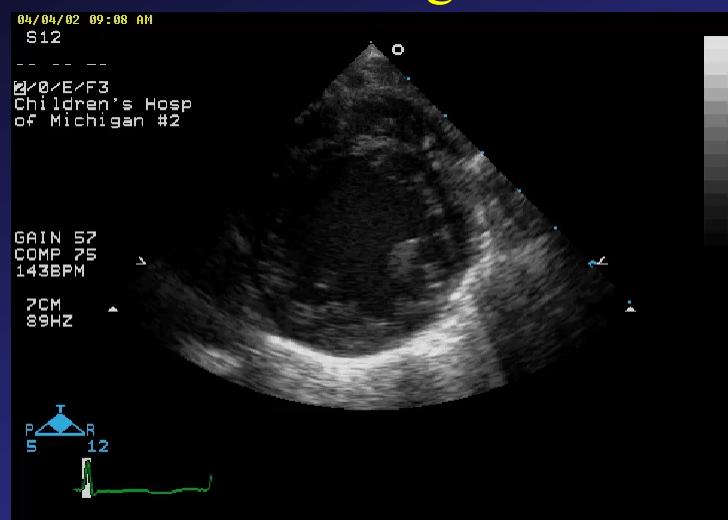
## Case 4 - Review



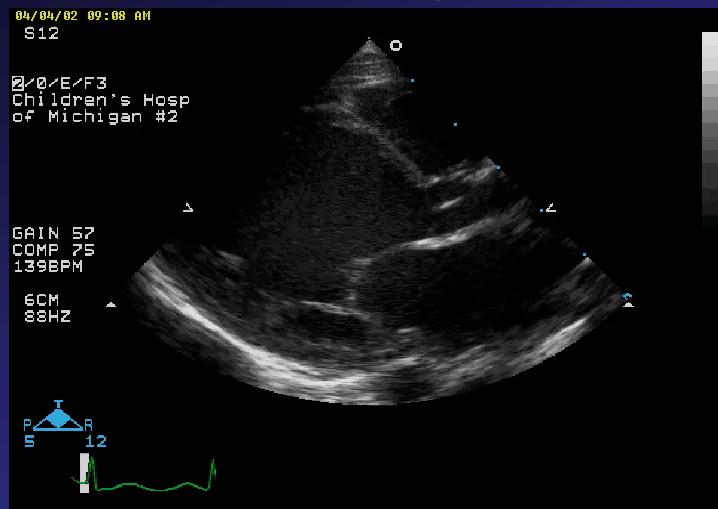
## Case 4 - Review



## Case 5 – Large PDA



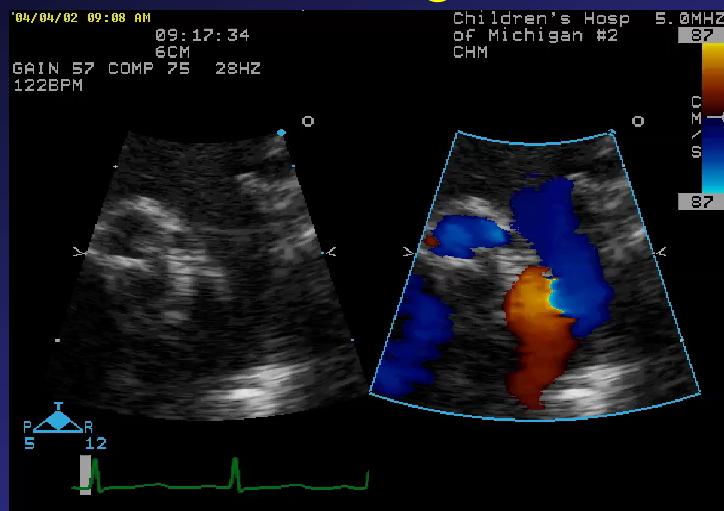
## Case 5 – Large PDA



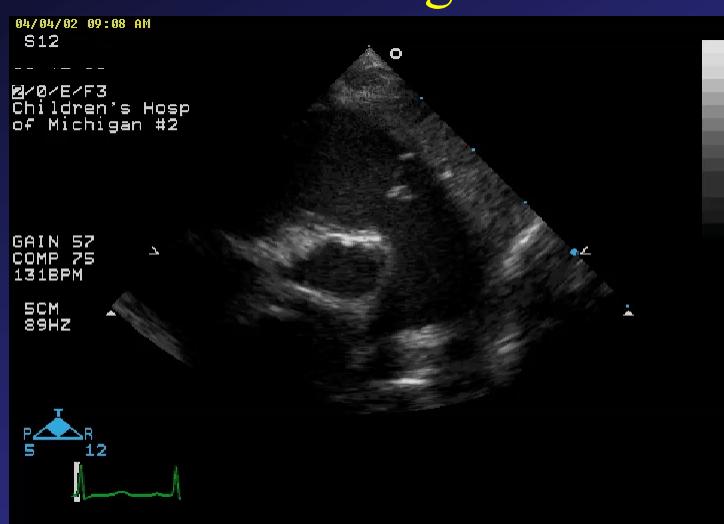
## Case 5 – Large PDA



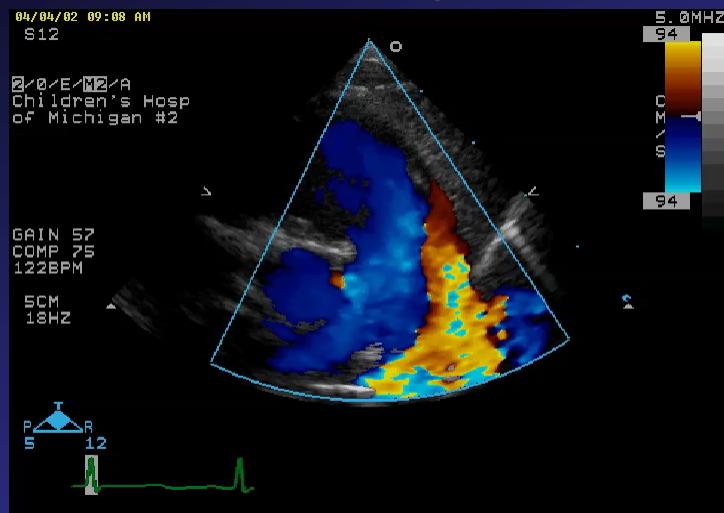
## Case 5 – Large PDA



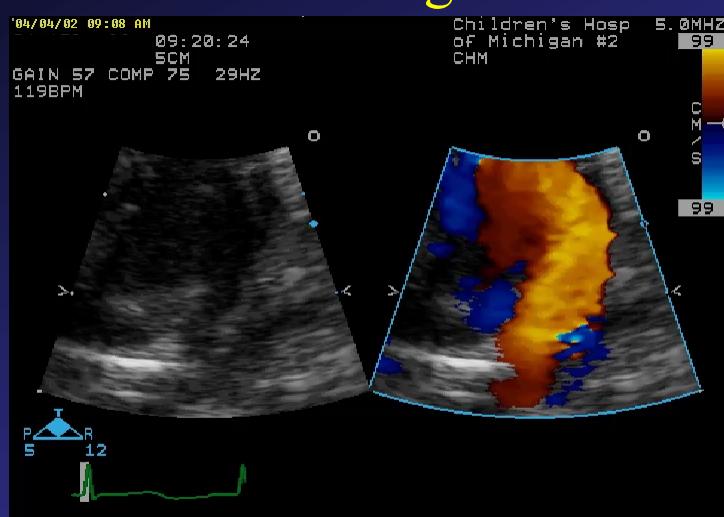
## Case 5 – Large PDA



## Case 5 – Large PDA

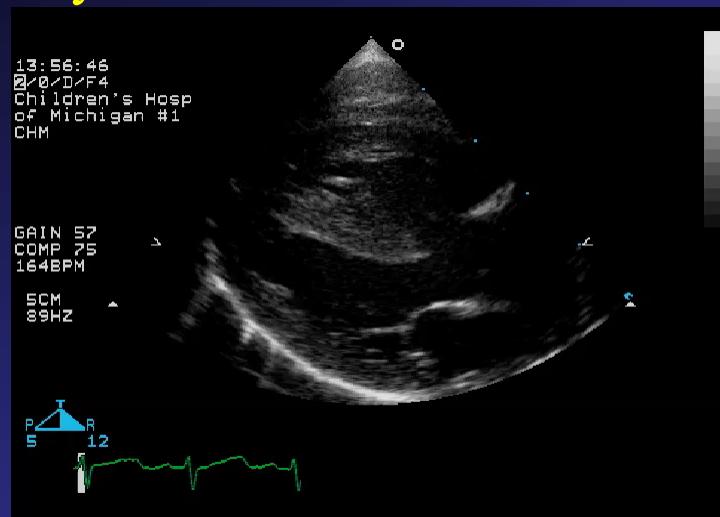


## Case 5 – Large PDA

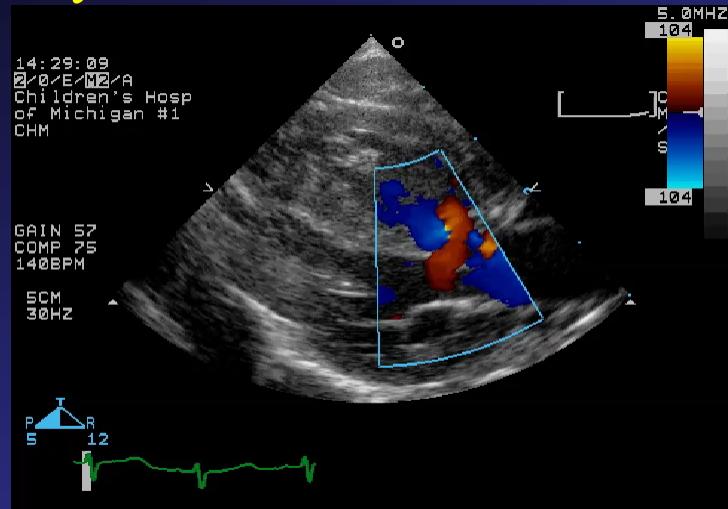




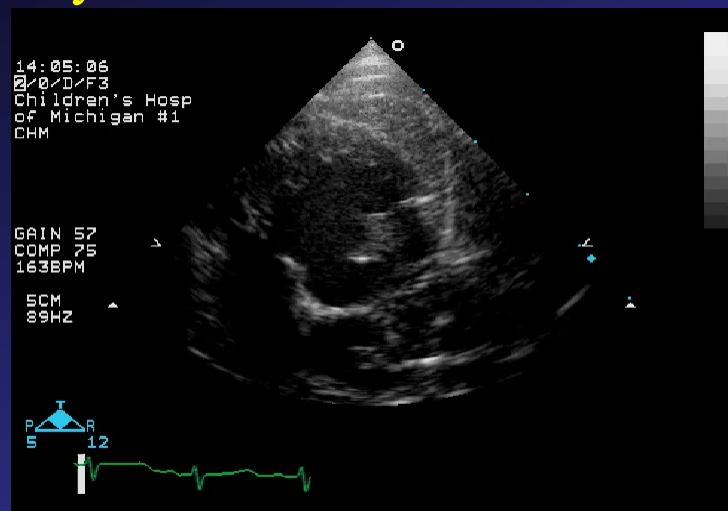
## Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur



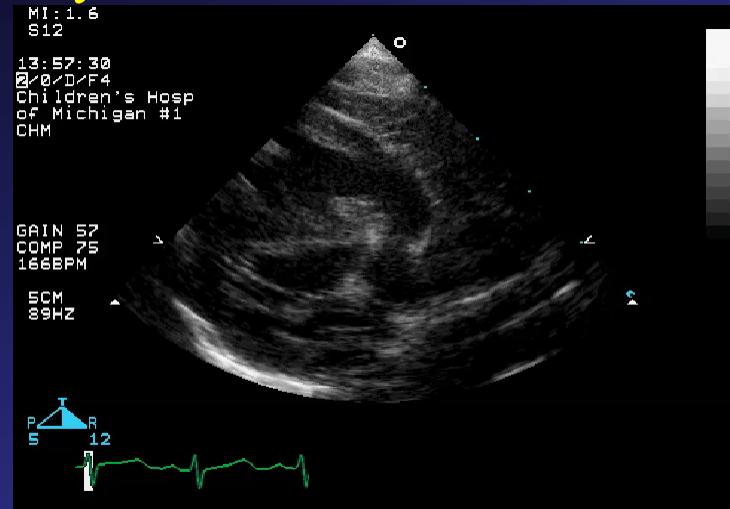
## Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur



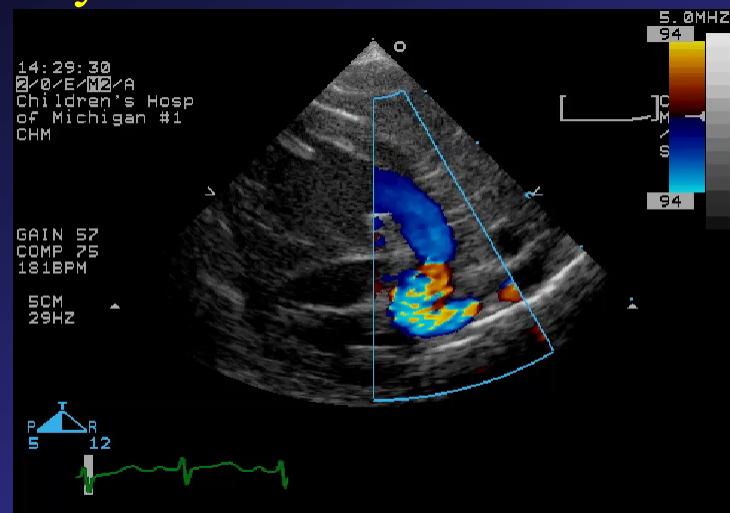
## Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur



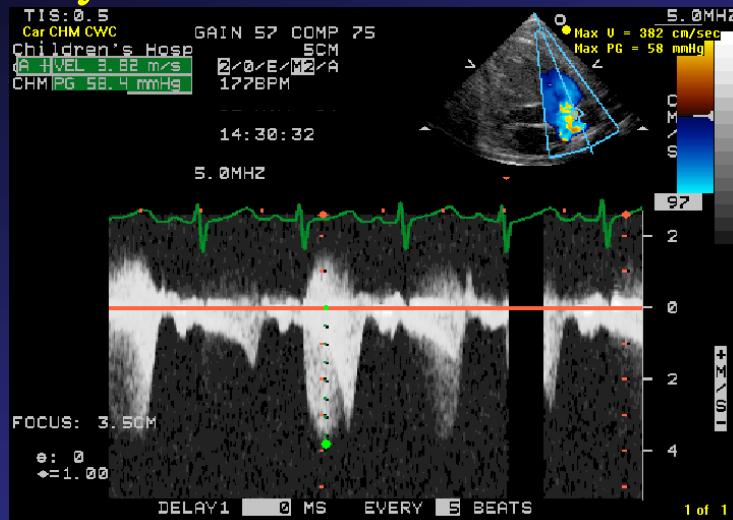
## Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur



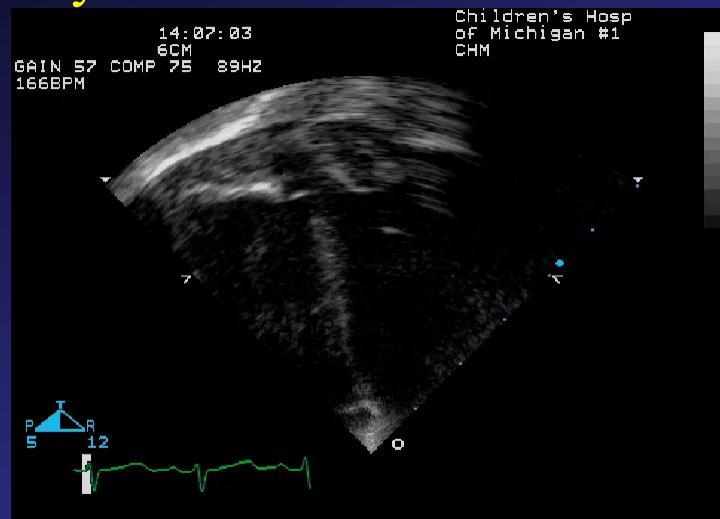
## Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur



## Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur



## Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur



## Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur



The defect shown in this example is:

1. Single ventricle
2. Transposition of the great arteries
3. Perimembranous VSD
- ★4. Tetralogy of Fallot
5. Complete atrioventricular canal

# Tetralogy of Fallot

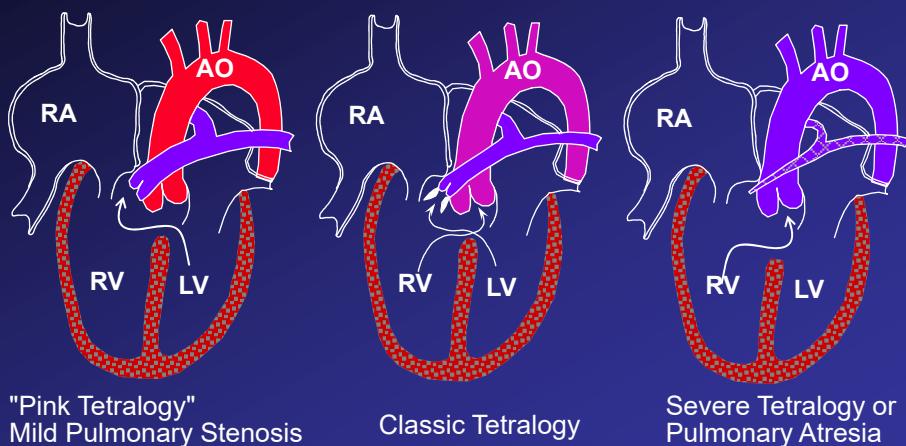
## Background

- Accounts for 10-13% of congenital heart disease
- Most common cyanotic CHD
- Usually present as asymptomatic murmur
  - Cyanosis usually develops/progresses with time
- Anatomy
  - Ventricular Septal Defect
  - Overriding Aorta
  - RV outflow obstruction
  - RV hypertrophy

Conal Septum  
Malalignment

# Tetralogy of Fallot

## Anatomic Variables



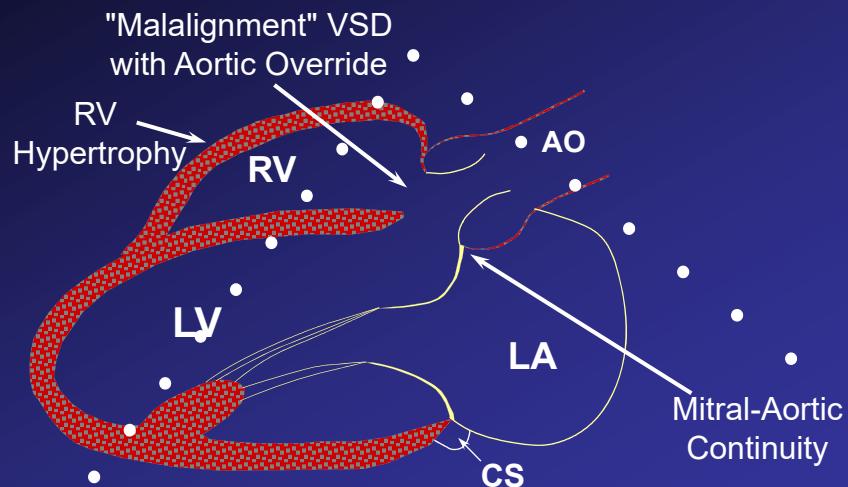
## Tetralogy of Fallot

### Associated Anomalies

- Valvular pulmonary stenosis - 50-60%
- Right aortic arch - 25%
- Atrial septal defect - 15%
- Coronary anomalies - 5%
- Muscular VSD - 2%

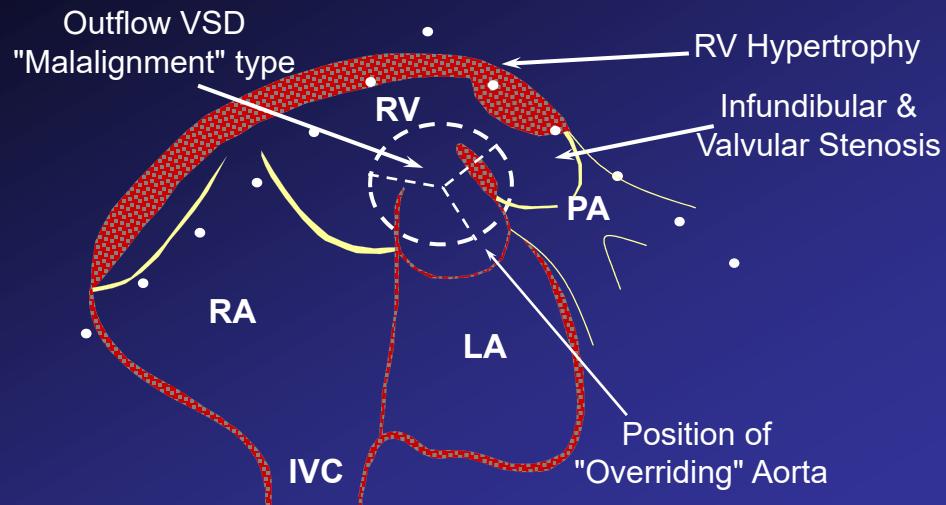
## Tetralogy of Fallot

### Parasternal Long Axis View



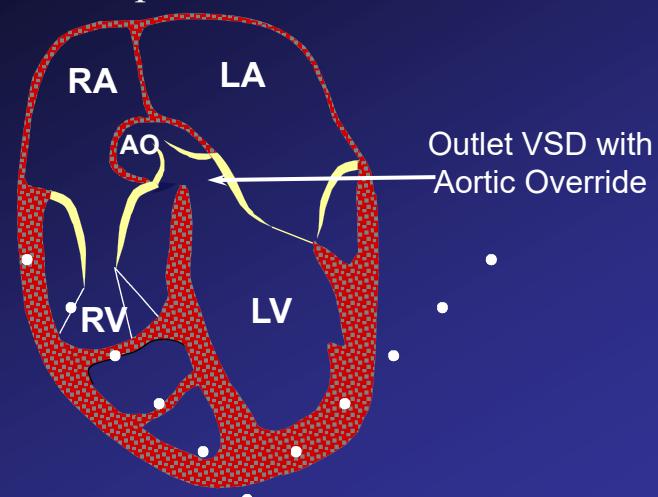
## Tetralogy of Fallot

### Parasternal Short Axis - Base



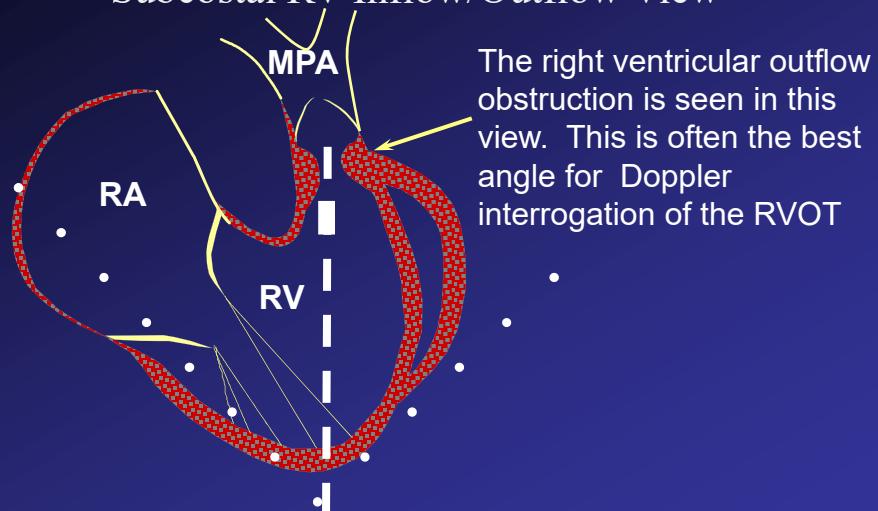
## Tetralogy of Fallot

### Apical Five-Chamber

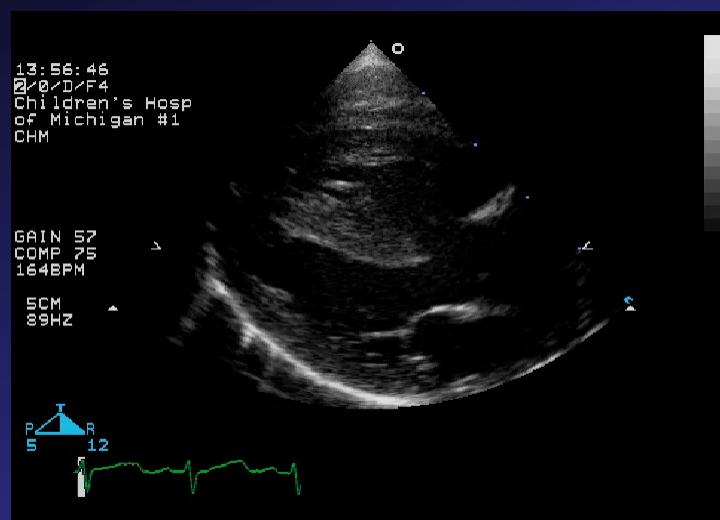


## Tetralogy of Fallot

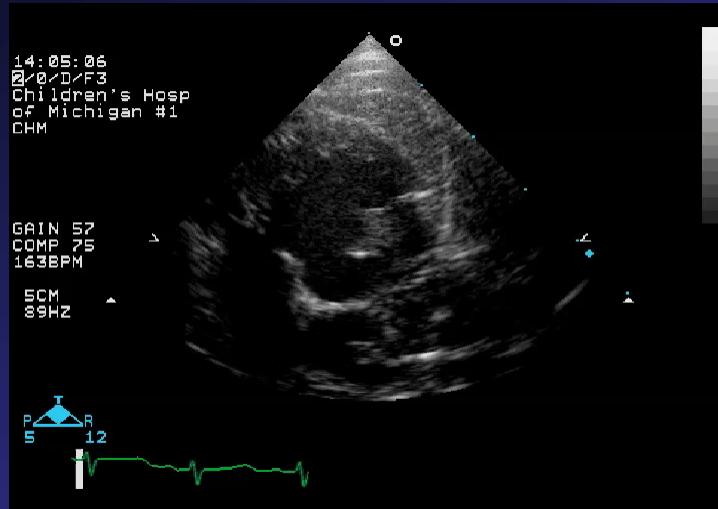
### Subcostal RV Inflow/Outflow View



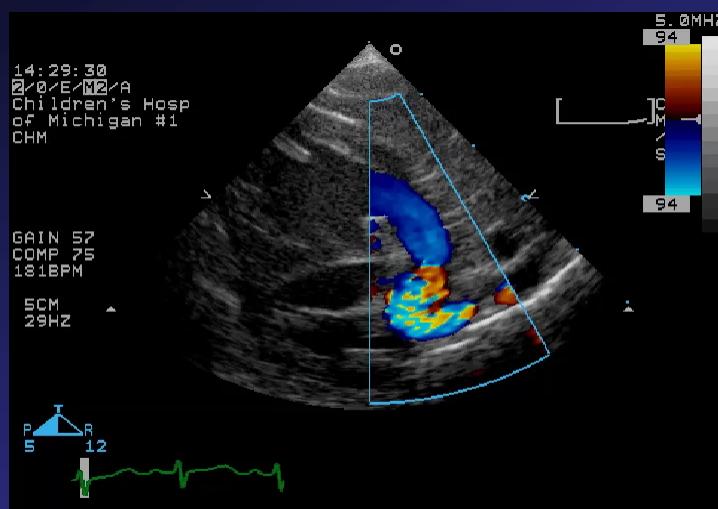
## Case 6 - Review



## Case 6 - Review



## Case 6 - Review

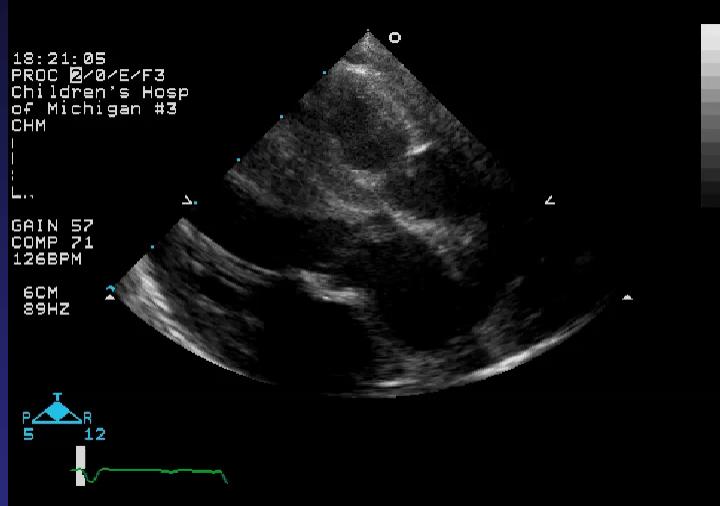


## Tetralogy of Fallot

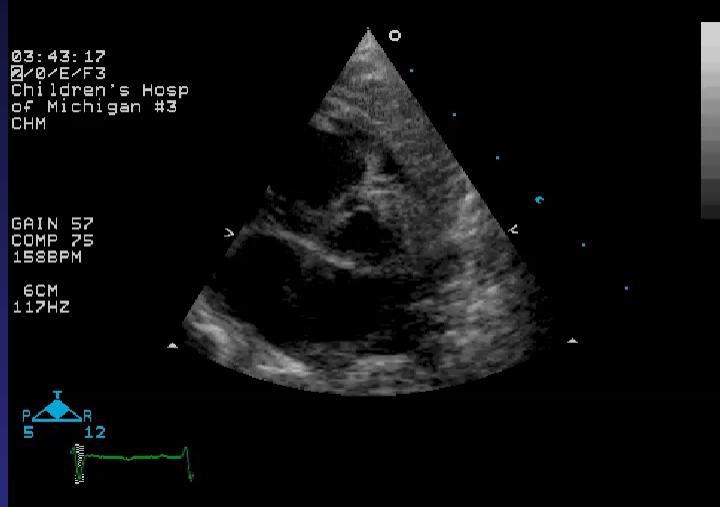
### Surgical Intervention

- Timing – usually during first 6 months
- VSD closure, relief of RVOTO obstruction
- Many repairs require a trans annular RV outflow patch with results in chronic severe pulmonary regurgitation
  - Likely need for late pulmonary valve replacement
- Rastelli type repair (VSD closure + RV to pulmonary artery conduit) may be required for complex anatomy – pulmonary atresia, coronary anomalies

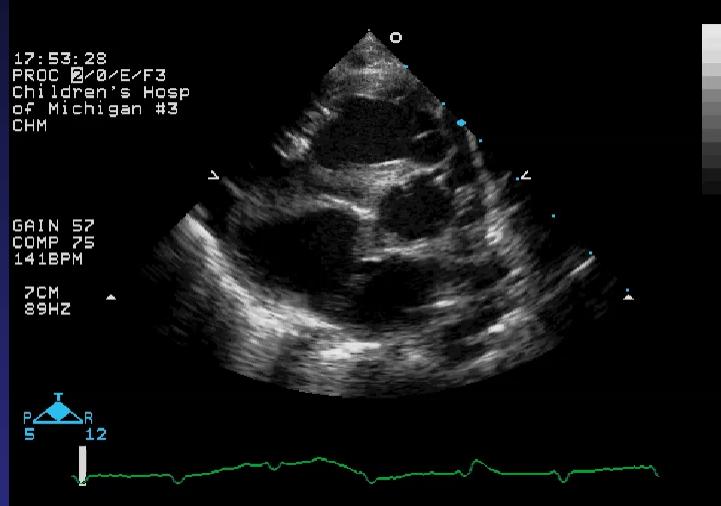
## Case 7 – 1 day old infant with tachypnea and SaO<sub>2</sub> of 76%



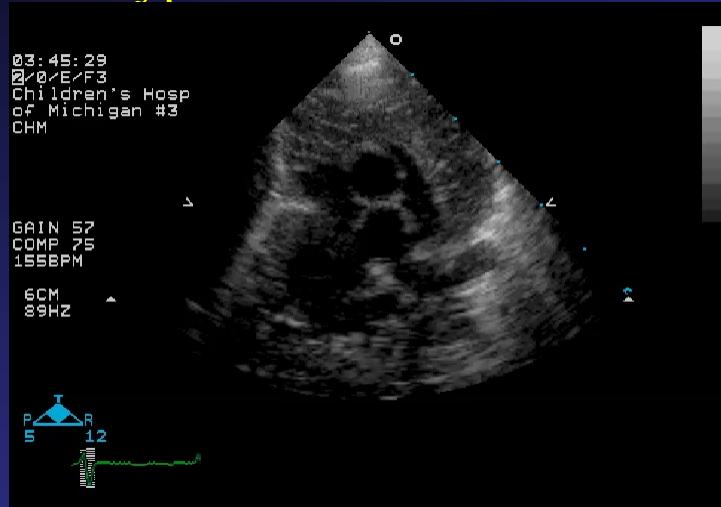
## Case 7 – 1 day old infant with tachypnea and SaO<sub>2</sub> of 76%



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## Case 7 – 1 day old infant with tachypnea and SaO<sub>2</sub> of 76%



What congenital heart defect is shown:

1. Perimembranous VSD
2. Truncus arteriosus
3. Corrected transposition of the great arteries (L-TGA)
- ★ 4. Complete transposition of the great arteries (D-TGA)
5. Tetralogy of Fallot

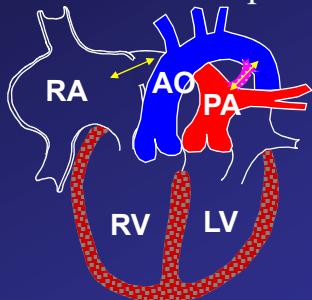
Which of the following is the preferred surgical palliation of this defect?

1. Rastelli operation
2. Mustard operation
- ★ 3. Jatene operation
4. Konno operation
5. Fontan operation

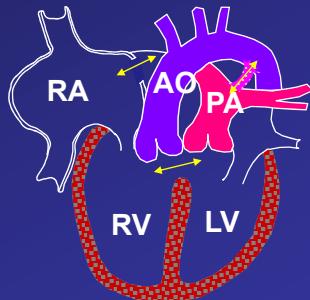
## D-Transposition of the Great Arteries

- Most common cyanotic CHD presenting in the newborn nursery
- 4-8% of CHD
- Very high mortality without intervention (90% at 1 year of life)

D-TGA Intact Septum



D-TGA w/ VSD



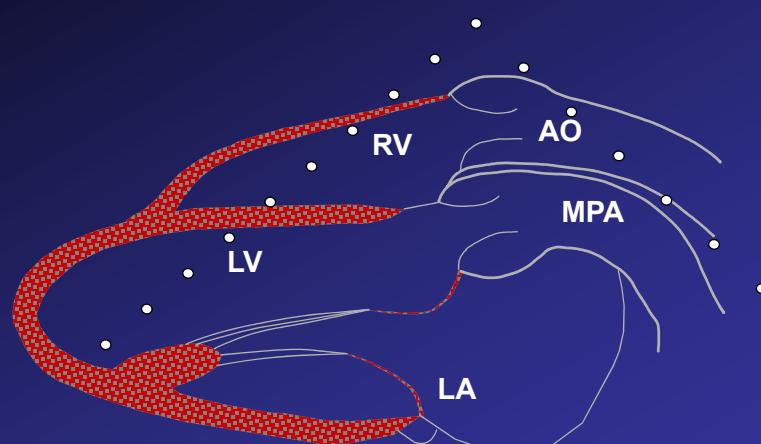
## D-Transposition of the Great Arteries

### Associated Anomalies

- VSD 40-45%
- Coronary anomalies ~ 40%
- Pulmonary stenosis (valve or sub valve) - 25%
- ASD
- PDA
- Coarctation - 5%

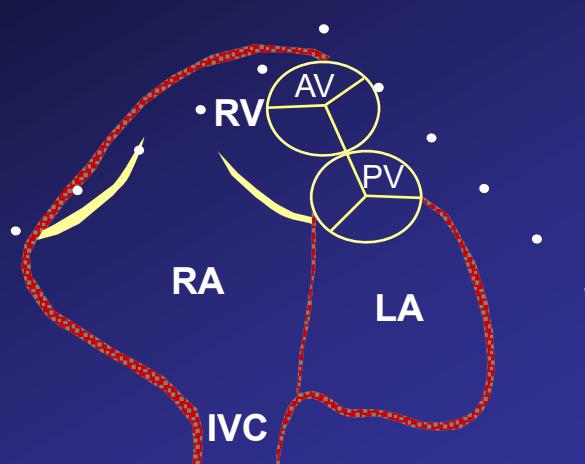
## D-Transposition of the Great Arteries

Parasternal Long Axis View



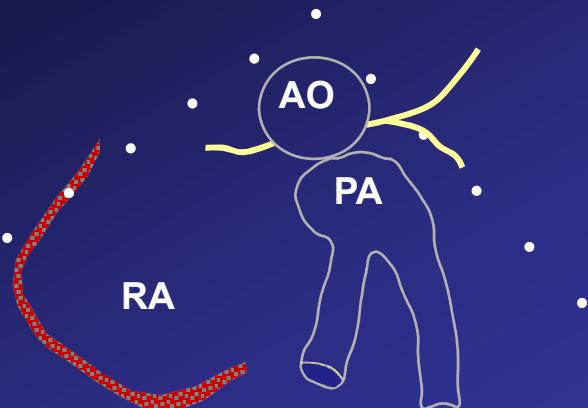
## D-Transposition of the Great Arteries

Parasternal Short Axis - Base

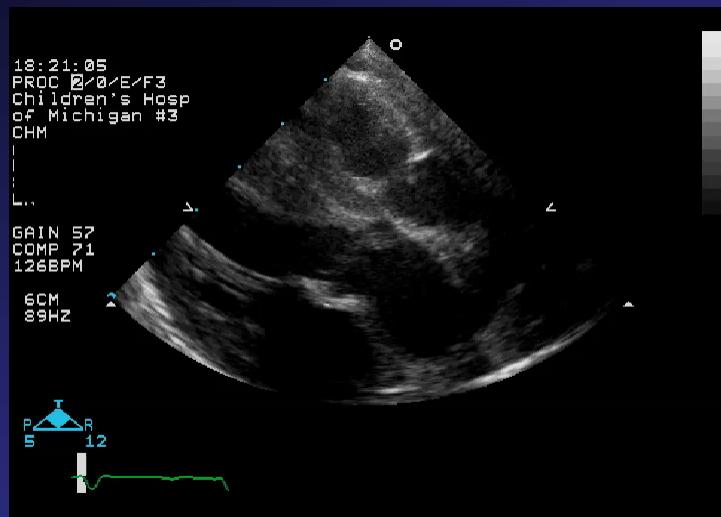


## D-Transposition of the Great Arteries

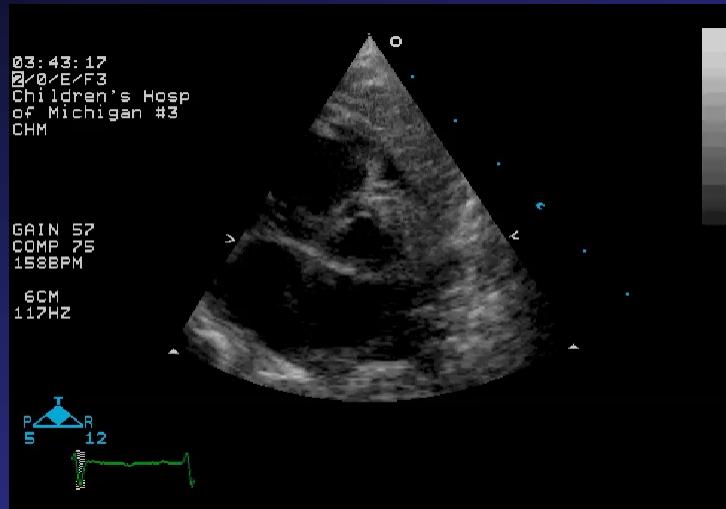
### High Parasternal Short Axis - Base



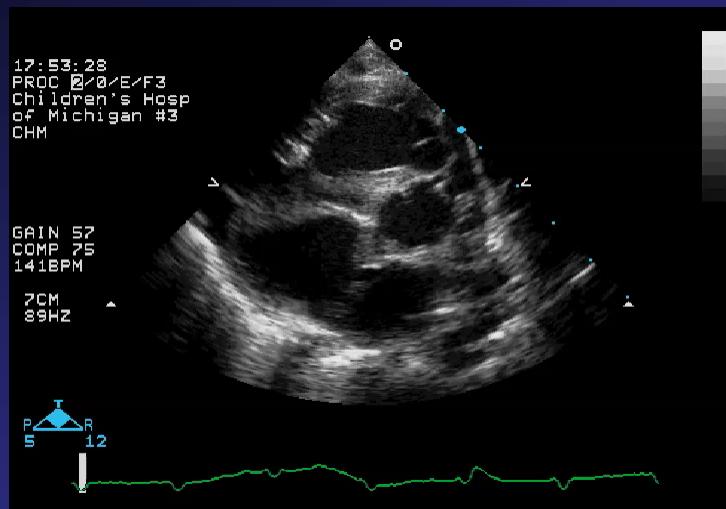
## Case 7 – Review



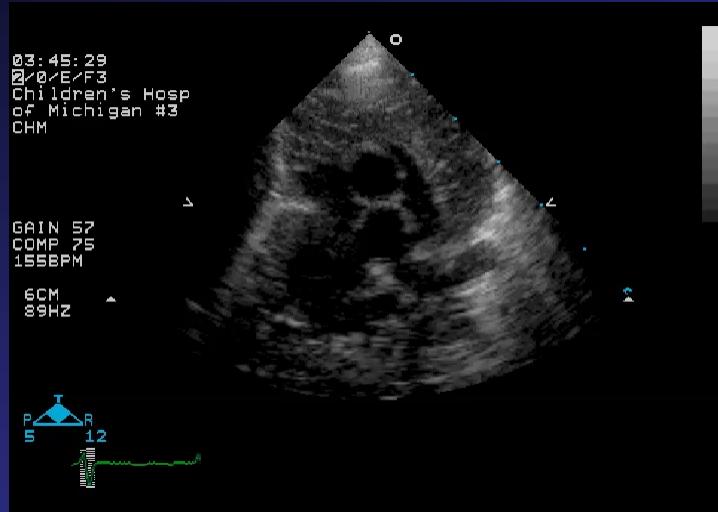
## Case 7 – Review



## Case 7 – Review



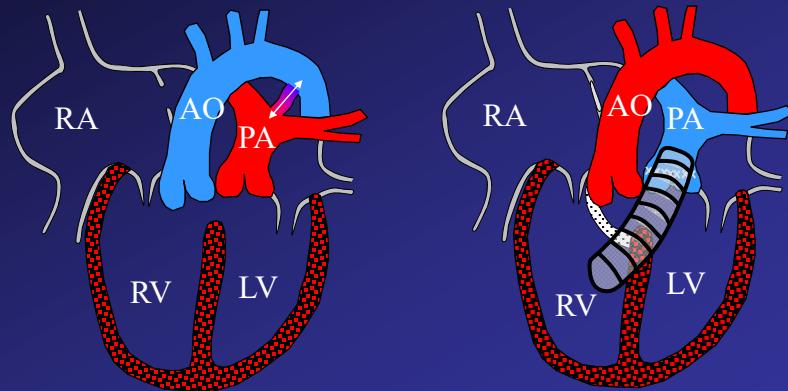
## Case 7 – Review



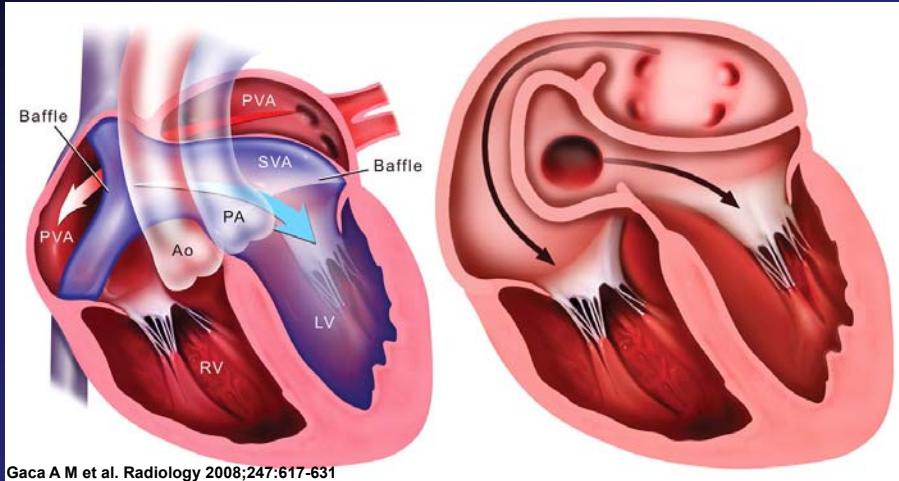
### D-Transposition of the Great Arteries Interventions

- Atrial balloon septostomy (Rashkind)
  - ♥ Increases saturation by improving atrial mixing
- Arterial switch (Jatene)
  - ♥ Performed within the first 1-2 weeks of life
- Atrial switch (Mustard/Senning)
  - ♥ Has been largely abandoned
- VSD closure/ RV-PA conduit (Rastelli)
  - ♥ Performed within the first few months of life
  - ♥ Used in the setting of d-TGA with PS/sub-PS
  - ♥ Requires conduit replacement/ future surgery

## D-TGA Intervention-Rastelli Procedure



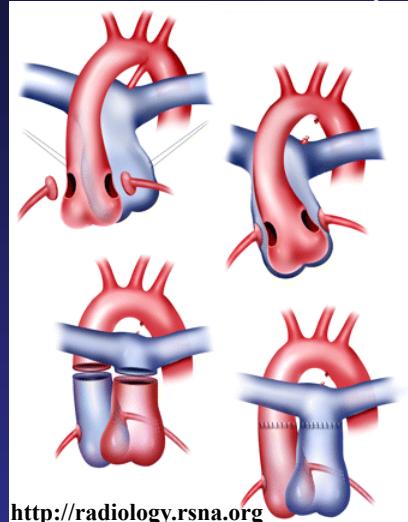
## D-TGA Atrial Switch (Mustard/Senning) Operation



Gaca A M et al. Radiology 2008;247:617-631

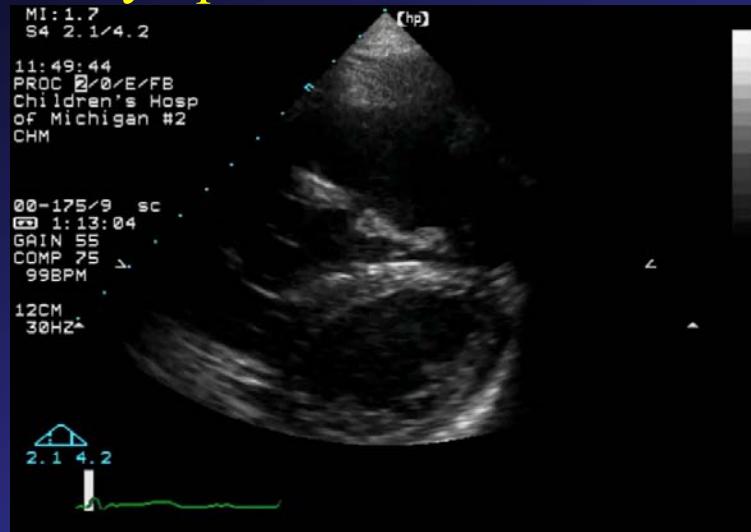
# TGA

## Jatene Arterial Switch Operation

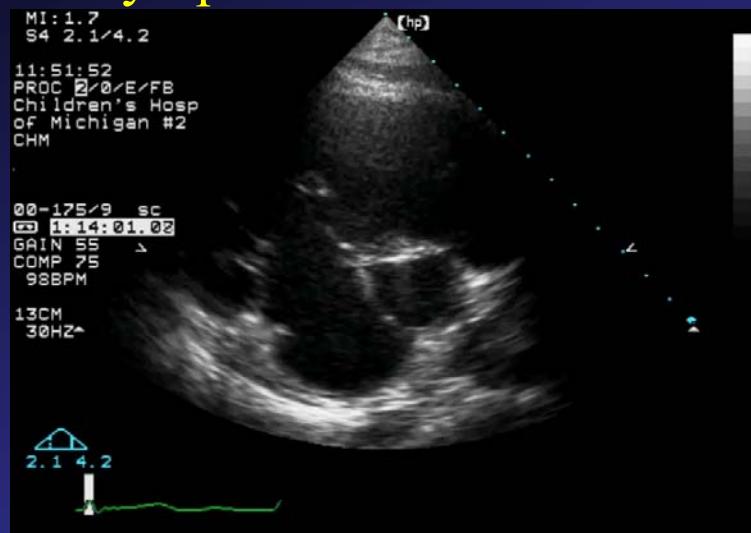


<http://radiology.rsna.org>

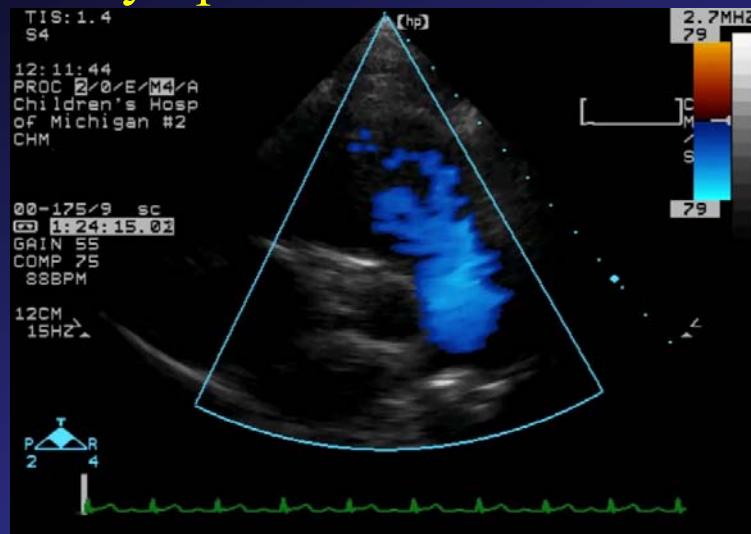
## Case 8 – 12 year old with asymptomatic murmur



## Case 8 – 12 year old with asymptomatic murmur



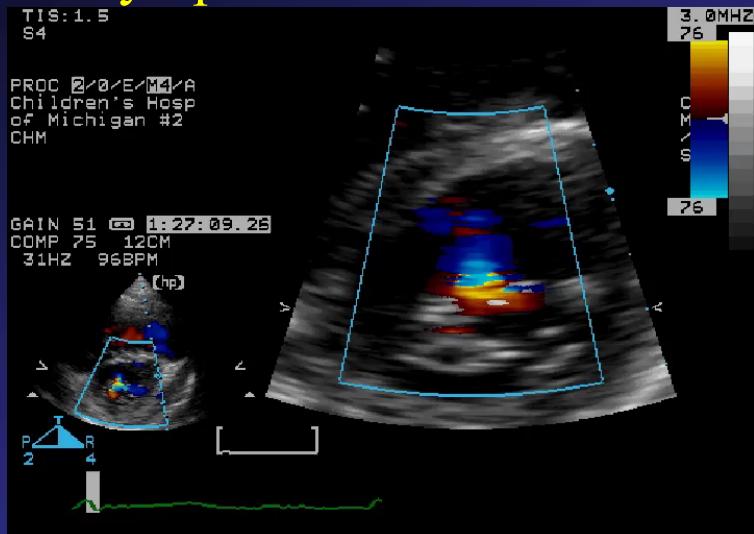
## Case 8 – 12 year old with asymptomatic murmur



## Case 8 – 12 year old with asymptomatic murmur



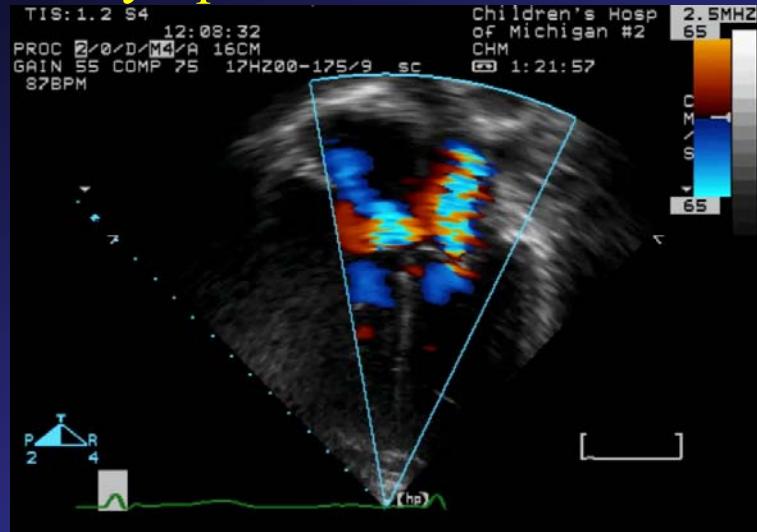
## Case 8 – 12 year old with asymptomatic murmur



## Case 8 – 12 year old with asymptomatic murmur



## Case 8 – 12 year old with asymptomatic murmur



The defect shown in this example is:

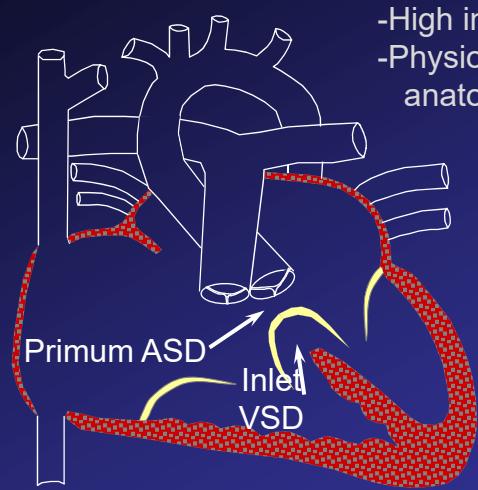
1. Secundum ASD
2. Sinus Venosus ASD
3. Perimembranous ASD
- ★ 4. Primum ASD
5. Coronary sinus ASD

A common associated defect with this anomaly, shown in this case, is:

1. Bicuspid aortic valve
2. Perimembranous VSD
3. Patent ductus arteriosus
4. Coarctation of the aorta
- ★ 5. Cleft mitral valve

## Atrioventricular Septal Defects

-3-5% of CHD  
-High incidence in Down Syndrome  
-Physiology depends on which anatomic defects are present



### Complete AVSD

1. Primum ASD
2. Inlet VSD
3. Common AV Valve

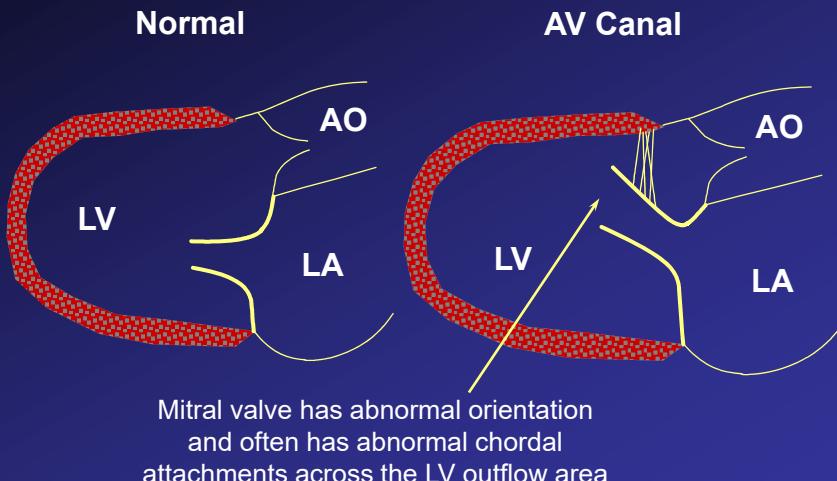
### Partial AVSD

1. Primum ASD
2. No VSD
3. Cleft Mitral Valve

# Complete Atrioventricular Canal Associated Anomalies

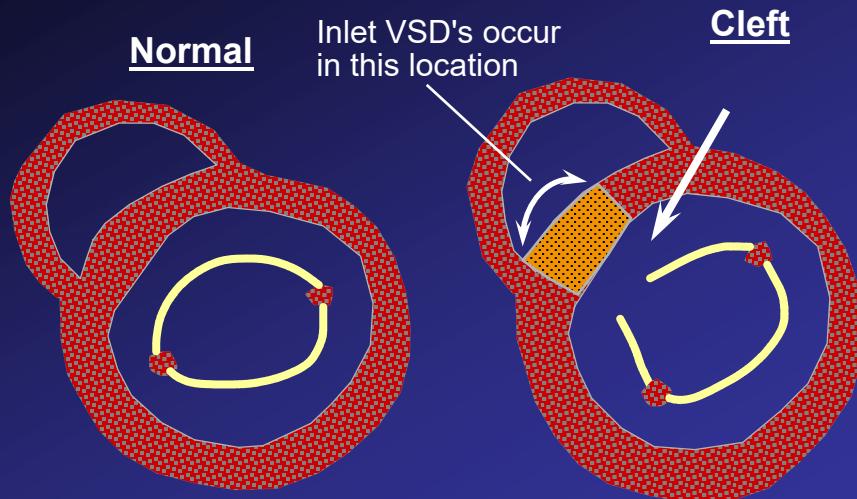
- Patent ductus arteriosus
  - Hypoplasia of one ventricle
  - AV valve problems - regurgitation
  - LVOT obstruction

## Atrioventricular Canal Long Axis View



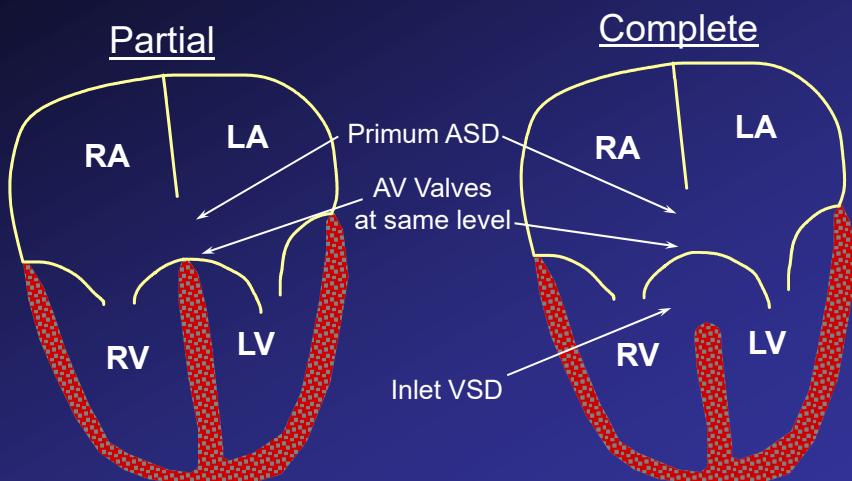
## Cleft Mitral Valve

Parasternal Short-Axis View



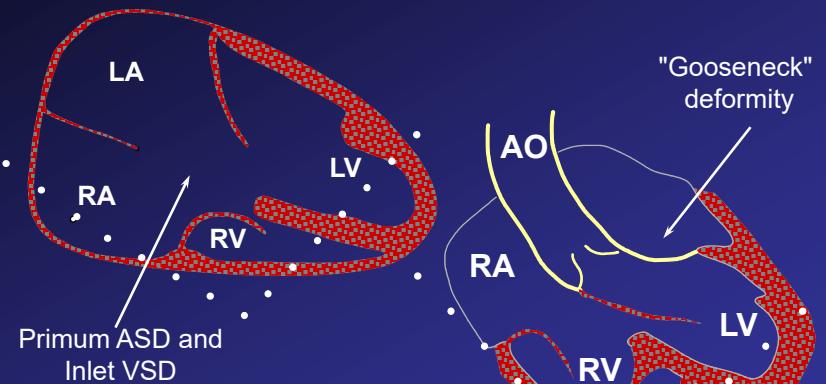
## Atrioventricular Canal

Apical 4-Chamber View



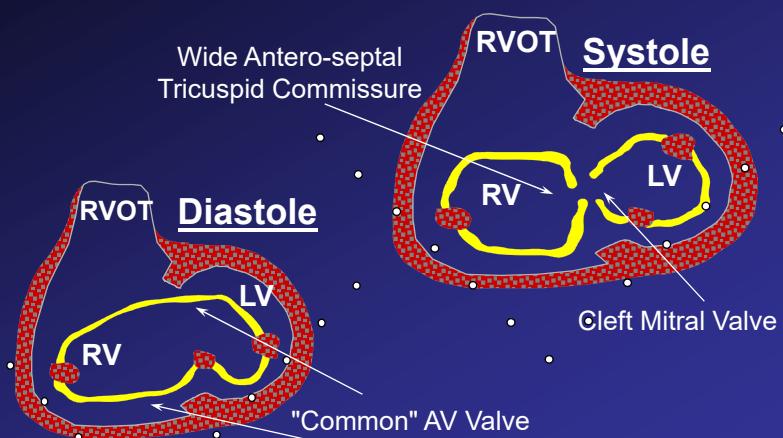
## Atrioventricular Canal Defects

### Subcostal Views

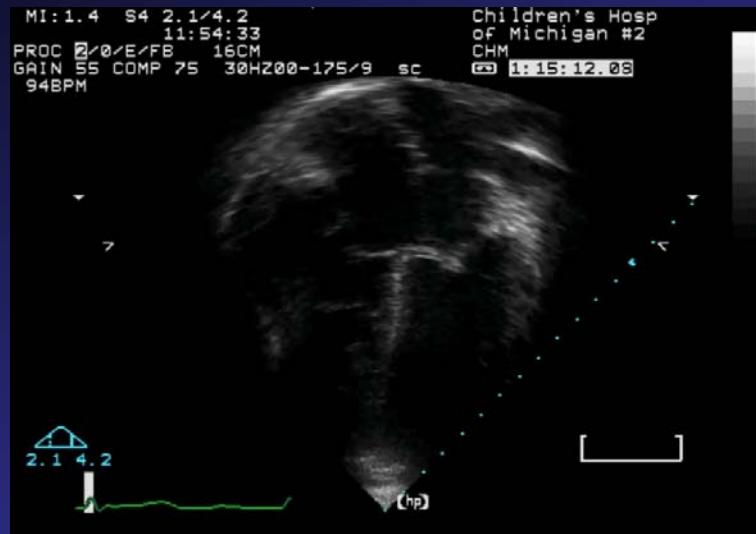


## Atrioventricular Canal Defects

### Subcostal Short Axis Views



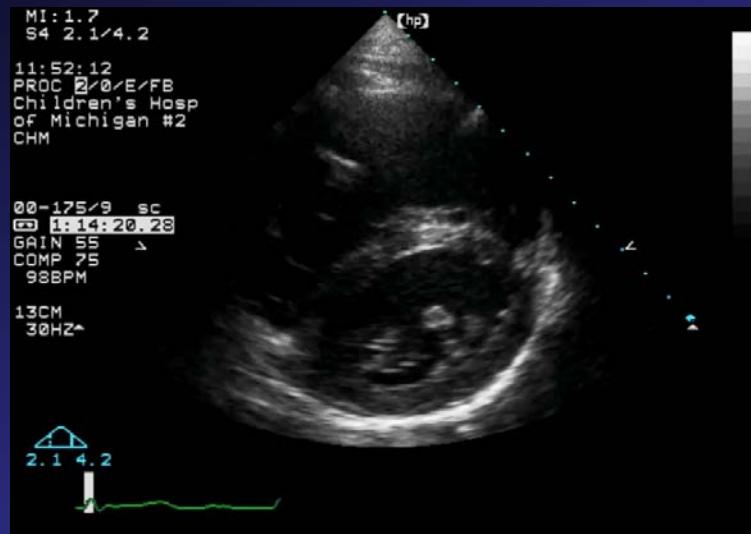
## Case 8 – Review



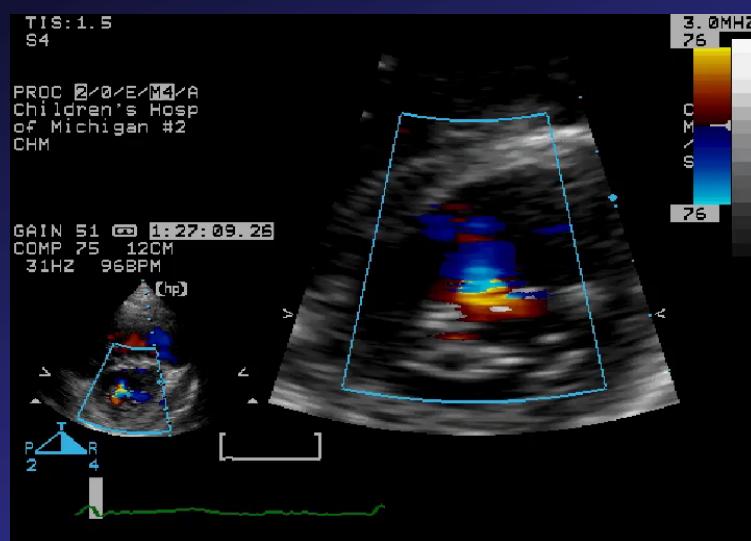
## Case 8 – Review



## Case 8 – Review



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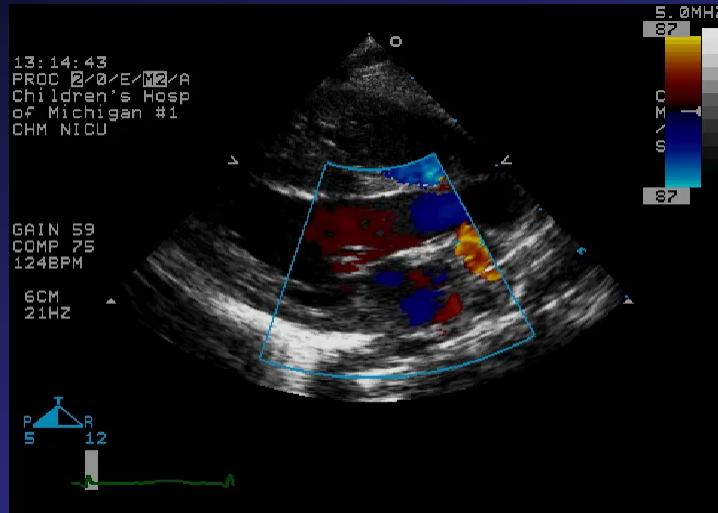




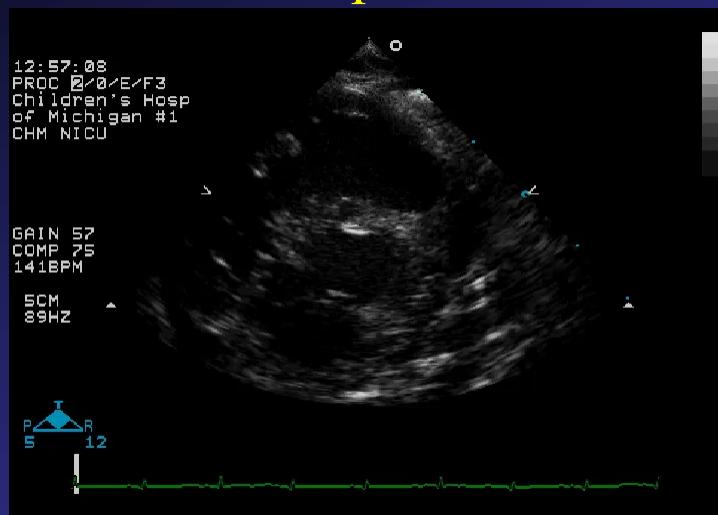
## Case 9 – Complete AV Canal



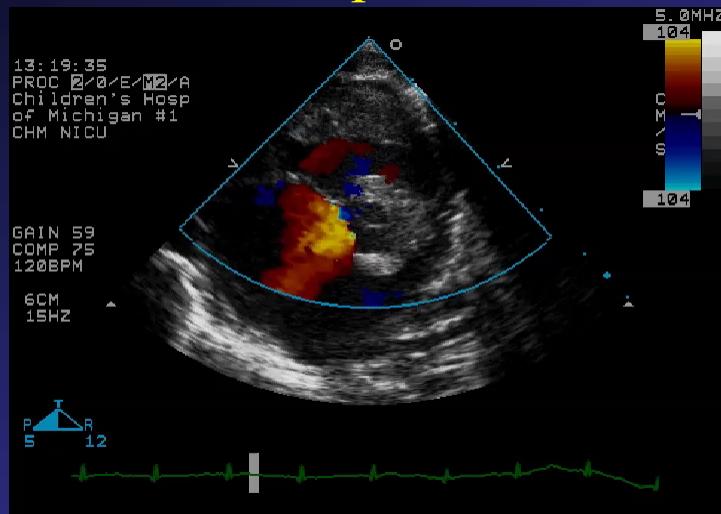
## Case 9 – Complete AV Canal



## Case 9 – Complete AV Canal



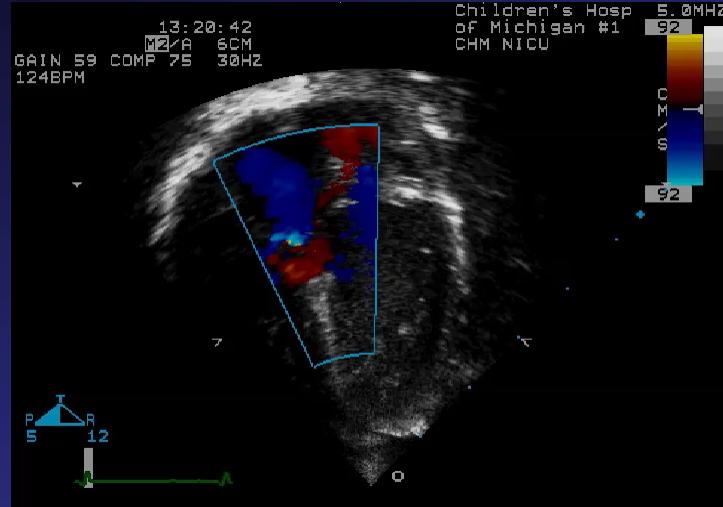
## Case 9 – Complete AV Canal



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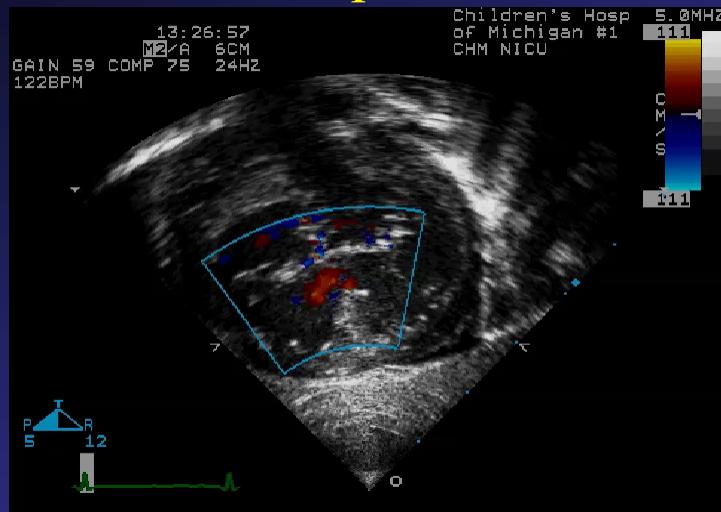
## Case 9 – Complete AV Canal



## Case 9 – Complete AV Canal



## Case 9 – Complete AV Canal



## AV Septal Defects

### Physiology

- Physiology dependent on which components of AV septal defect are present
- If 1° ASD and no VSD - physiology similar to isolated ASD (right sided volume overload)
- Complete AVSD - marked volume and pressure overload (VSD shunt physiology)
- AV valve regurgitation may exacerbate volume overload and symptoms of heart failure

## AV Septal Defects

### Surgical Intervention

- Partial AVSD
  - Usually electively repaired at age 2-4 years
  - Complicating features (AVV regurgitation, LVOTO) may necessitate earlier intervention
- Complete AVSD
  - Usually repaired by 6 months of age (earlier in trisomy 21) to prevent pulmonary vascular obstructive disease

## AV Septal Defects

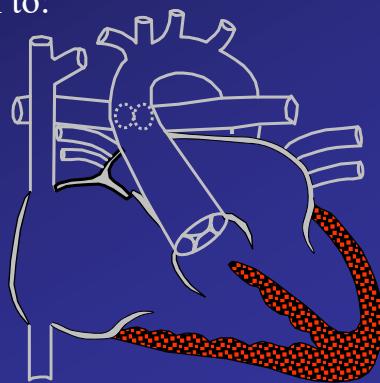
### Outcomes After Surgery

- Surgical outcomes
  - 3-4% operative mortality for complete AVSD
  - <1% operative mortality for partial AVSD
- Late reoperation in 10-15%
  - Left AV valve regurgitation
  - Left ventricular outflow tract obstruction
    - ♥ Hypoplasia of the outflow tract
    - ♥ Accessory AV valve tissue
    - ♥ Discrete subaortic membrane

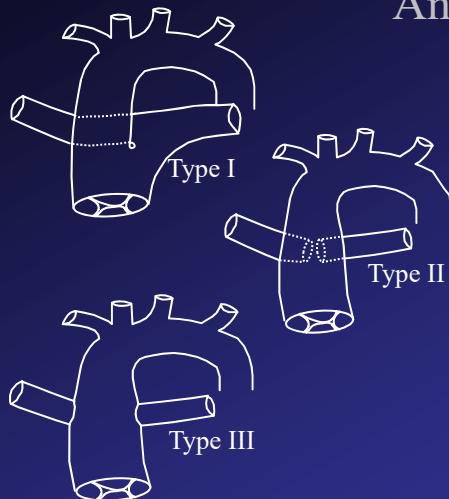
## Truncus Arteriosus

### Anatomy

- Characteristic anatomy characterized by:
  - Single arterial vessel that arises from the base of the heart and gives origin to:
    - ♥ Systemic arteries
    - ♥ Pulmonary arteries
    - ♥ Coronary arteries
  - Single semilunar valve



## Truncus Arteriosus Anatomy



### Associated Defects

- Abnormal coronaries (37-49%)
- Right aortic arch (30%)
- Abnormal truncal valve
- Absent pulmonary artery (16%)
- Interrupted aortic arch (15%)
- Left SVC (12%)
- Secundum ASD (9-20%)

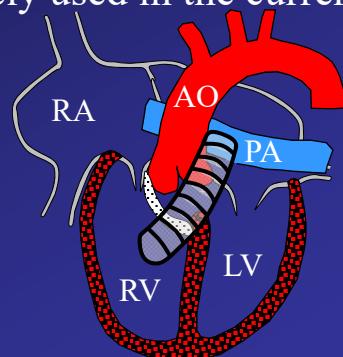
## Truncus Arteriosus Clinical Aspects

- Patients usually present due to the presence of a cardiac murmur
- Complete mixing of systemic and pulmonary venous blood results in cyanosis
- Excessive pulmonary blood flow causes sign and symptoms of congestive heart failure
- The cyanosis is generally mild

## Truncus Arteriosus

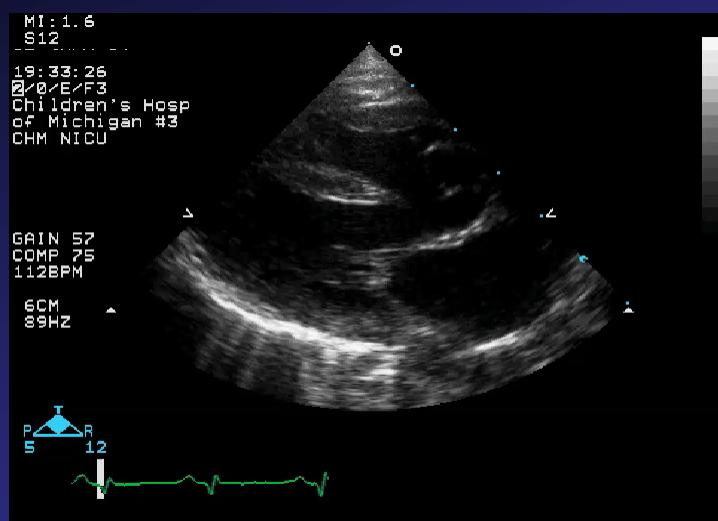
### Treatment

- Requires surgical repair in the first weeks of life
- Median sternotomy requiring bypass
- Palliative PA band rarely used in the current era
- Rastelli type repair
  - Close VSD to truncus
  - Disconnect PAs
  - RV to PA conduit



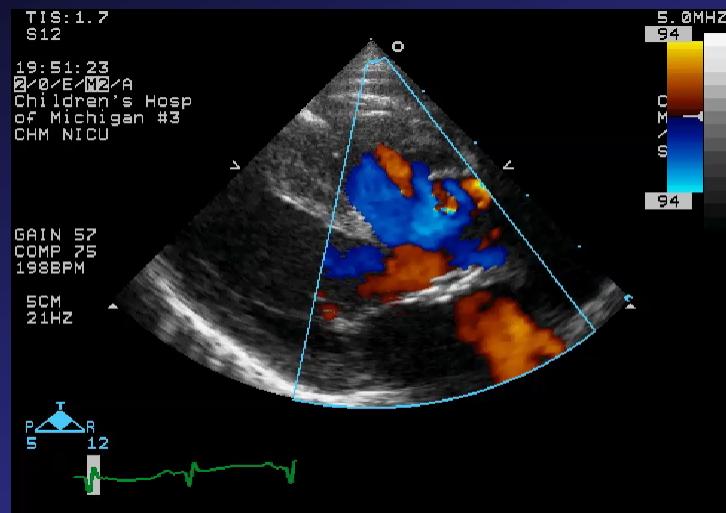
## Case 10

### Truncus Arteriosus



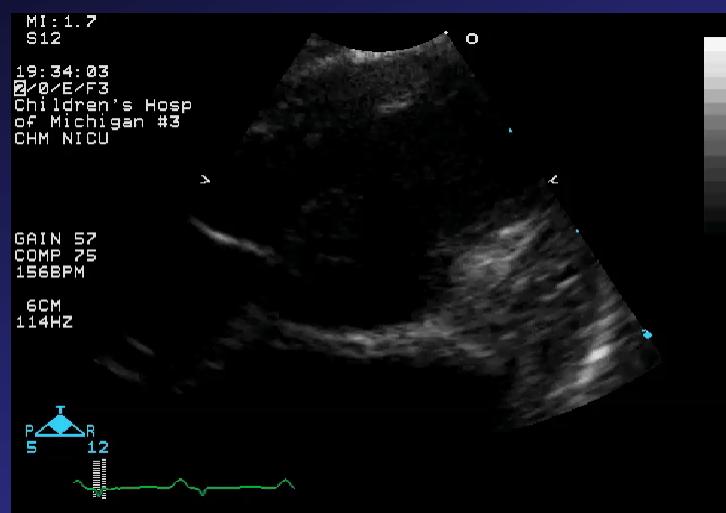
## Case 10

### Truncus Arteriosus



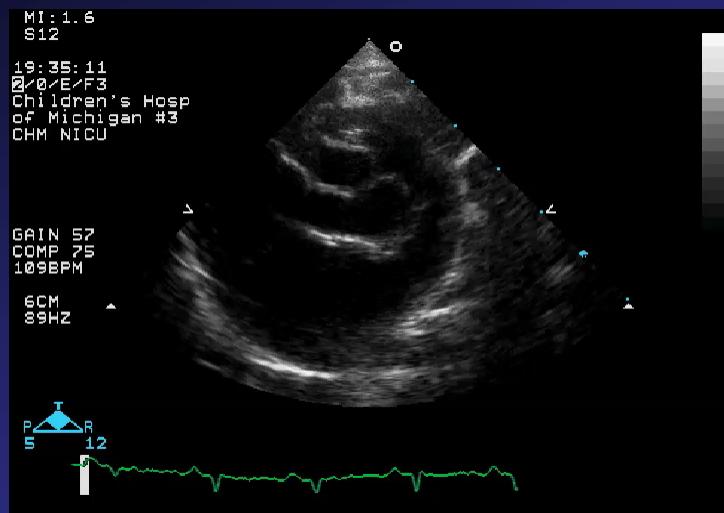
## Case 10

### Truncus Arteriosus



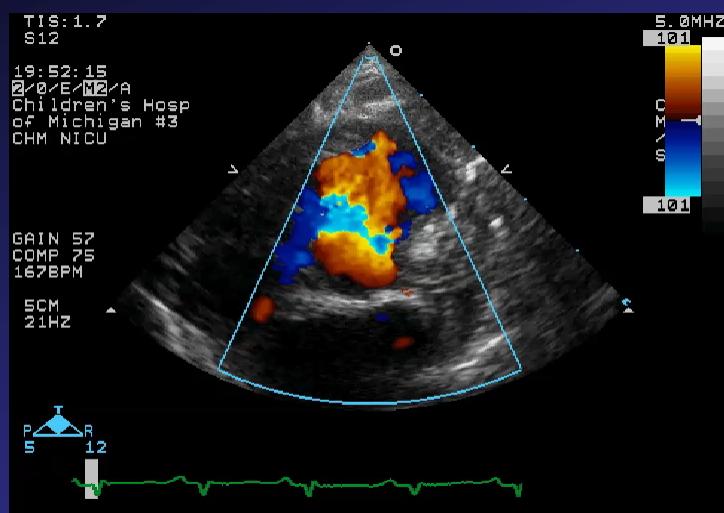
## Case 10

### Truncus Arteriosus



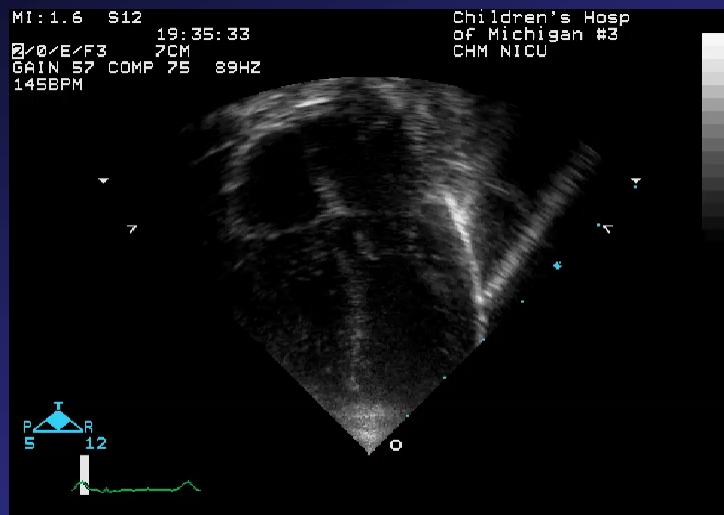
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### Truncus Arteriosus



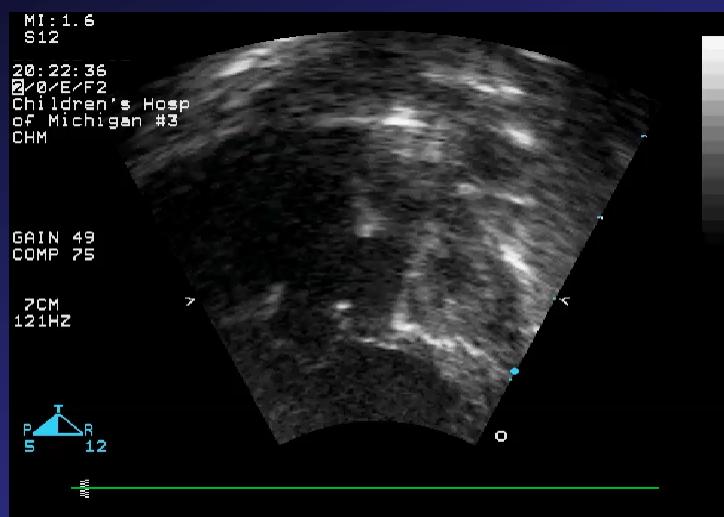
## Case 10

### Truncus Arteriosus



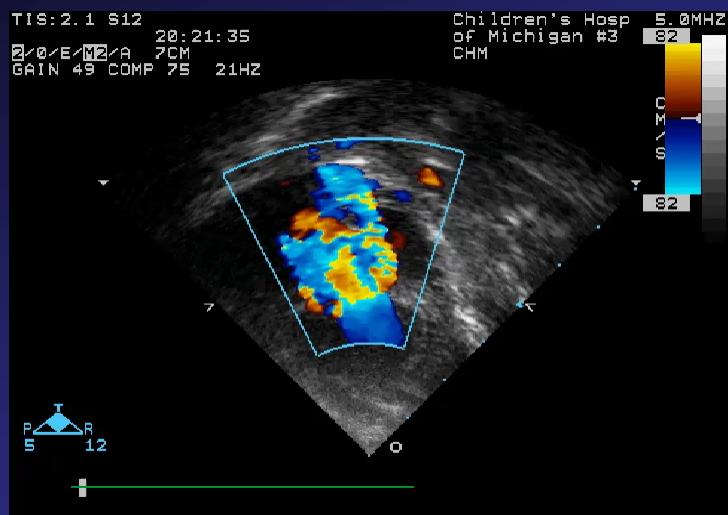
## Case 10

### Truncus Arteriosus



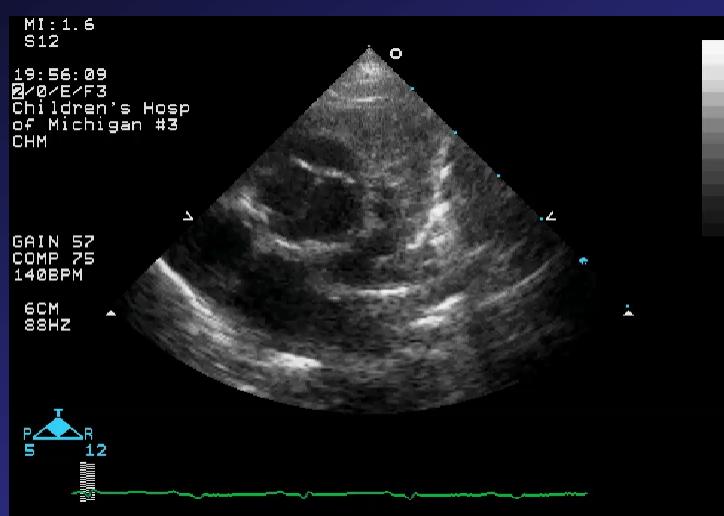
## Case 10

### Truncus Arteriosus



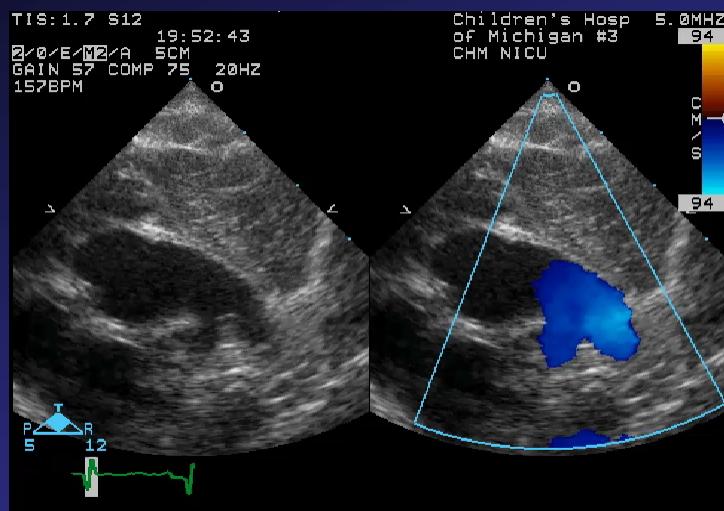
## Case 10

### Truncus Arteriosus

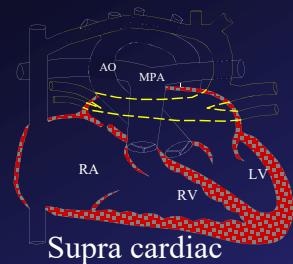


## Case 10

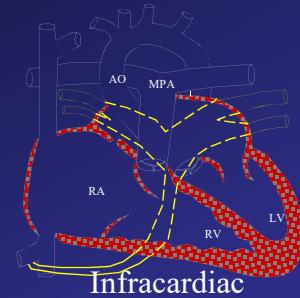
### Truncus Arteriosus



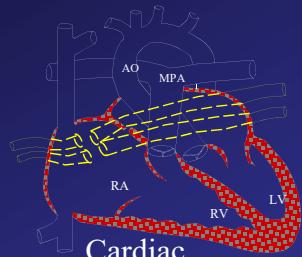
## Total Anomalous Pulmonary Venous Return Anatomic Types



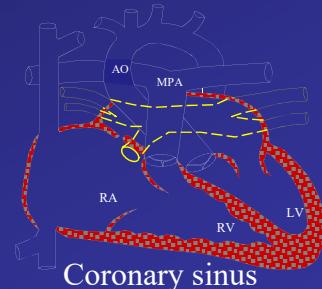
Supra cardiac



Infracardiac



Cardiac



Coronary sinus

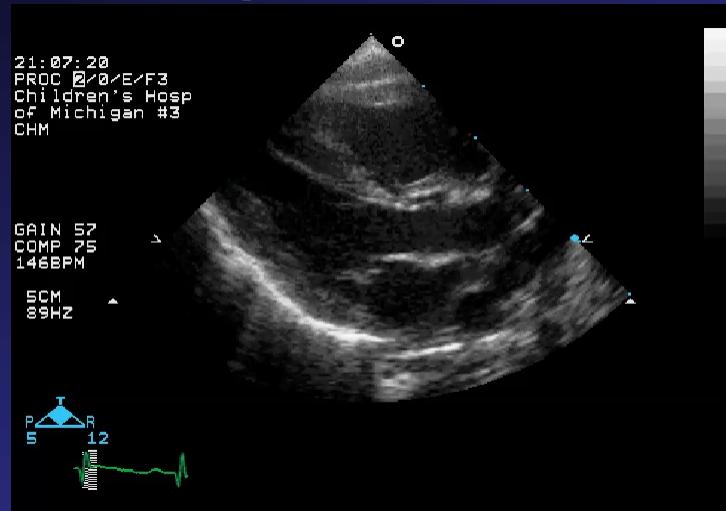
## Total Anomalous Pulmonary Venous Return

### Echo Clues

- Enlarged right heart
- Right to left atrial shunting
- Unusual “membranes” in left atrium
- Abnormal flow in systemic venous system
- Obstruction may occur at different levels
  - Most common - infracardiac
- May be remarkably asymptomatic (in absence of obstruction)

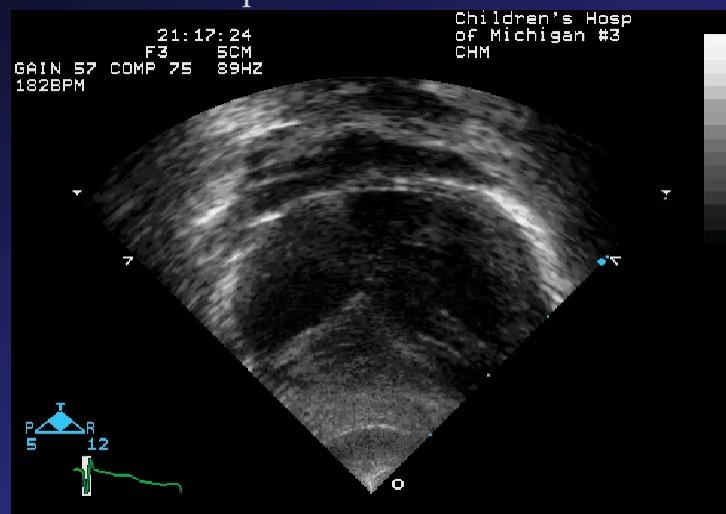
## Case 12

Supra cardiac TAPVR



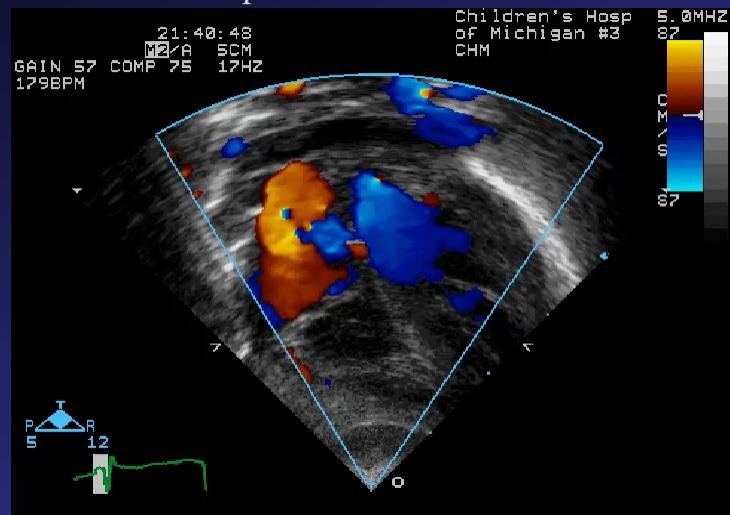
## Case 12

Supra cardiac TAPVR



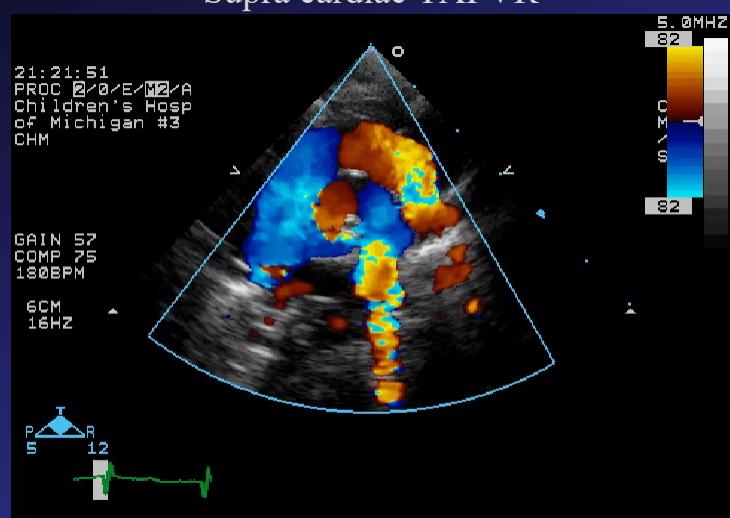
## Case 12

Supra cardiac TAPVR



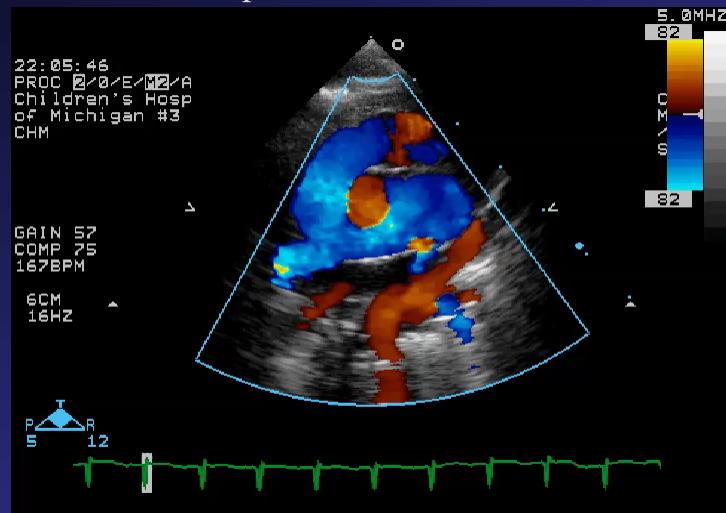
## Case 12

Supra cardiac TAPVR



## Case 12

Supra cardiac TAPVR



## Anomalous Left Coronary Artery

From the Pulmonary Artery - ALCAPA



- Rare congenital anomaly
- Usually presents at 2-3 m of age
- Results in severe LV ischemia
- Present as dilated CM, CHF
- ECG often diagnostic
- Patients survive w/ collateral flow
- Surgery done to re-implant vessel

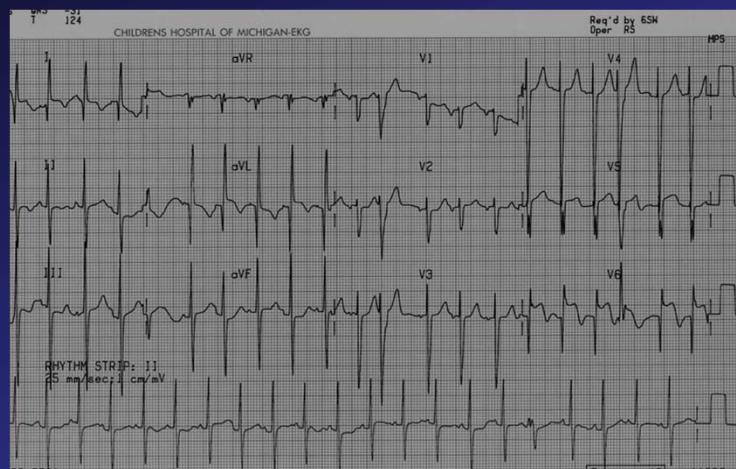
## Anomalous Left Coronary Artery

### Echocardiographic Clues

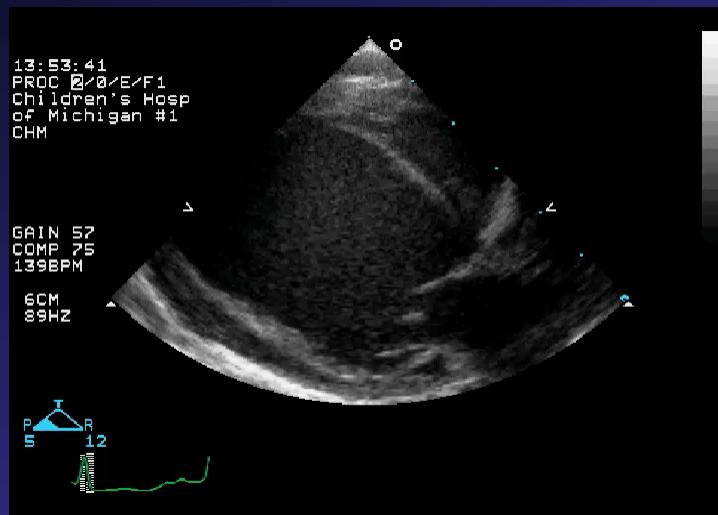
- Left ventricular dysfunction (usually severe)
- Mitral insufficiency – due to LV dilation/dysfunction, papillary muscle infarction
- Endocardial fibroelastosis of LV and/or papillary muscles
- Failure to identify proximal LCA from aorta
- Unusual flow into main pulmonary artery

# Anomalous Left Coronary Artery Artery

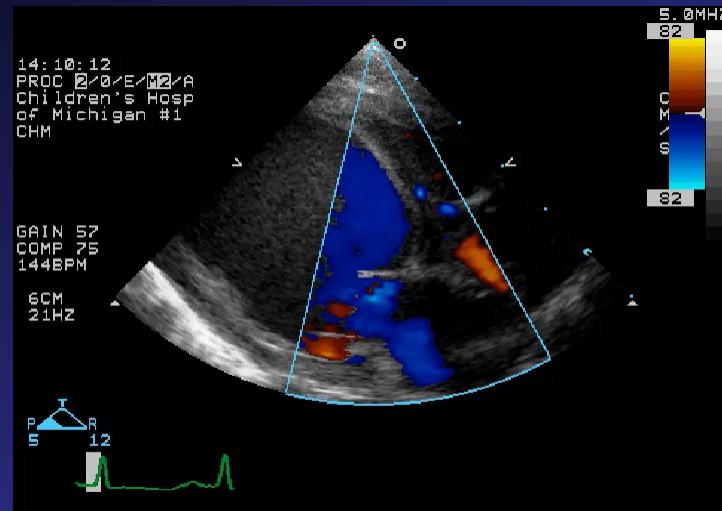
From the Pulmonary Artery - ALCAPA



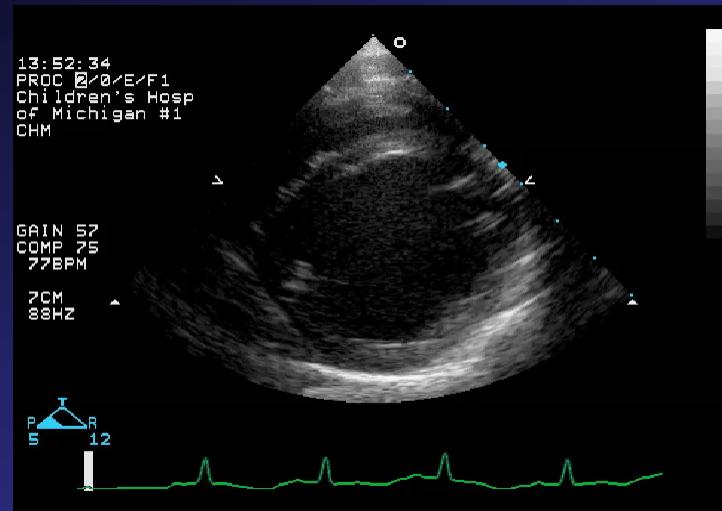
## Case 11 ALCAPA



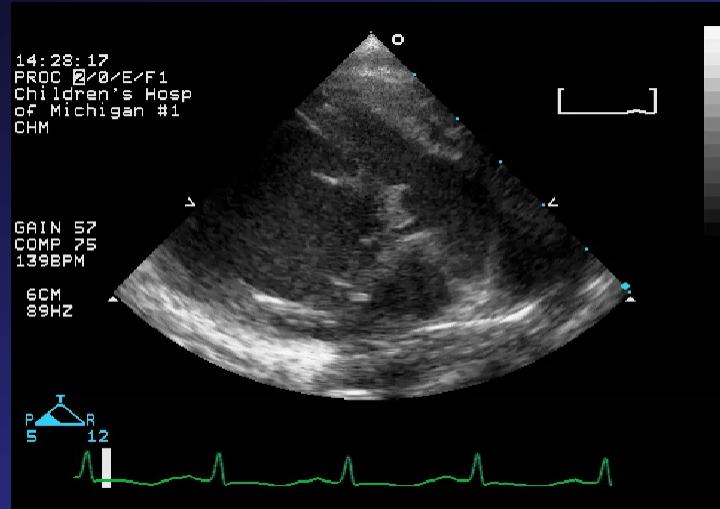
## Case 11 ALCAPA



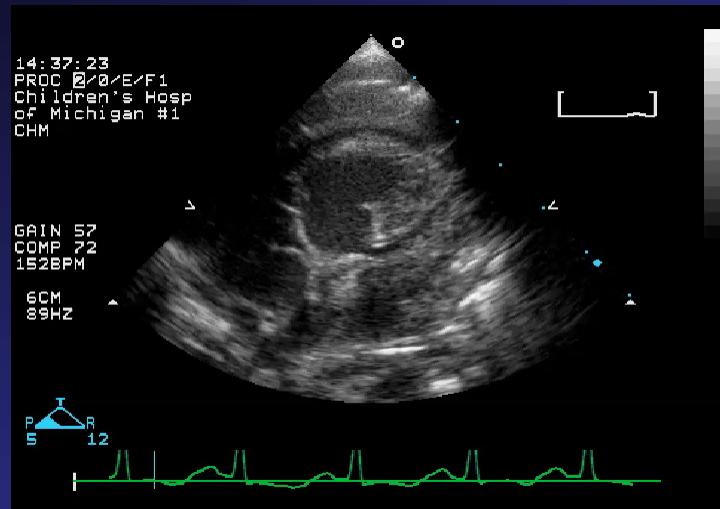
## Case 11 ALCAPA



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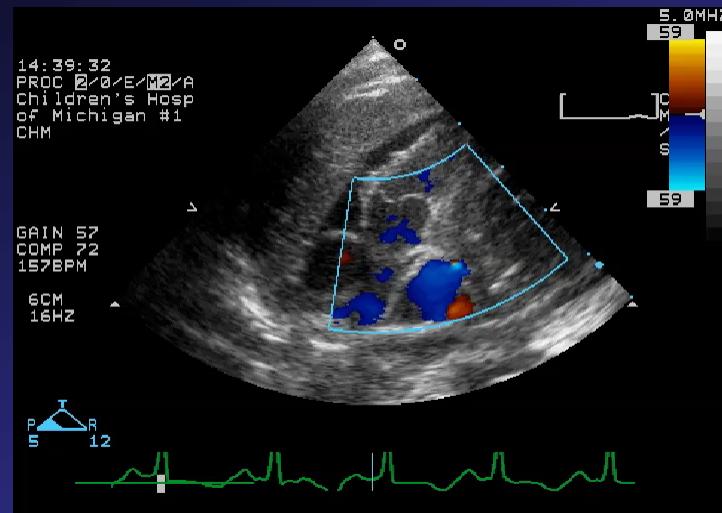


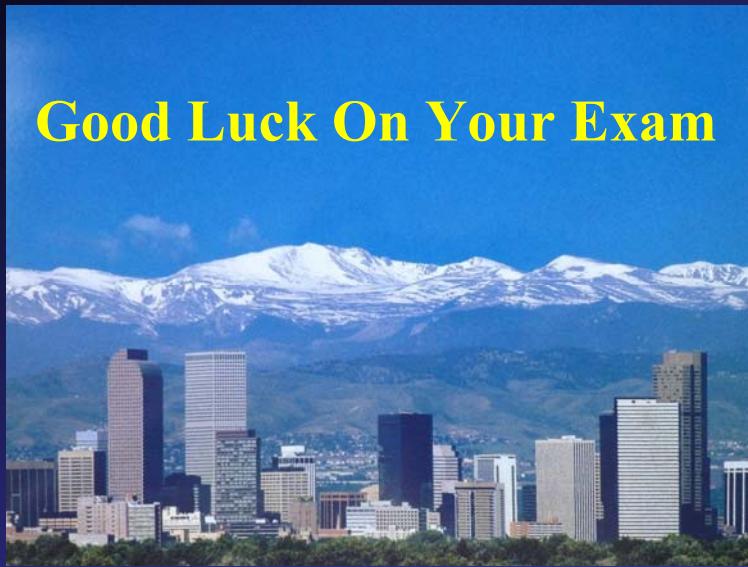
## Case 11 ALCAPA



## Case 11

### ALCAPA





**Good Luck On Your Exam**

**Questions?**  
[michael\\_pettersen@mednax.com](mailto:michael_pettersen@mednax.com)